ADAPTIVE FUNCTIONING AND FUNCTIONAL READING SKILLS

IN WILLIAMS SYNDROME

Submitted in partial fulfilment of

the requirements for the degree of

DOCTOR OF PSYCHOLOGY (CLINICAL NEUROPSYCHOLOGY)

Macquarie University

Gabrielle Brawn

2013

Abstract

This thesis investigated adaptive functioning or everyday living skills in Williams syndrome (WS) and considered the relationship between adaptive functioning with demographic and environment variables and functional reading skills. WS is a genetic disorder, resulting from a microdeletion on chromosome 7, with a prevalence of approximately 1 in 7,500 births. There are characteristic physical and behavioural features of WS and the intellectual and cognitive profile of the disorder has been studied extensively due to interesting peaks and valleys of ability. Less well understood is the profile of adaptive functioning in WS and the profile of reading abilities. This thesis comprises of a general introduction, three chapters and a conclusion. The introduction explains the conceptual links between the three main chapters from a design and research perspective. Chapter One is a systematic review of adaptive functioning in WS and includes 15 published studies and 3 Ph.D dissertations. The relationship between adaptive skills and demographic variables such as age, gender and intellect were considered. The review found the overall level of adaptive functioning to be low in WS; however, there was evidence of age related changes with adults demonstrating comparatively lower skills when compared to children and adolescents. Also, the pattern of relative strengths and weaknesses was different across these age spans. Methodological evaluation found that further research into adaptive functioning was warranted in WS, with updated assessment tools, further investigation of age-related differences, along with the inclusion of environmental factors which may contribute to adaptive functioning. The second chapter follows on from this review, with an empirical study that aimed to investigate adaptive functioning in WS across the age range of children through to adults using an up-to-date measure of adaptive functioning. Environmental contributions to adaptive functioning were also explored. Thirty individuals with a genetically confirmed diagnosis of WS, all with the common ~1.6Mb deletion participated in the study. Global adaptive functioning was considered, as were domain and subdomain scores. Group averages were explored, as well as individual profiles of strength and weakness. The study revealed significant variability in adaptive abilities, and while neither gender nor intellectual functioning were found to be related to adaptive functioning, chronological age and some aspects of the family environment were found to significantly relate to adaptive abilities. The third chapter investigated functional literacy abilities in WS to consider whether reading abilities were found to be related to adaptive functioning, in particular, independence in daily activities, such as telling the time, managing money, job skills etc. A cognitive neuropsychological approach was used to examine lexical and nonlexical reading abilities in WS to determine whether there was any evidence of different reading patterns, along with cognitive and intellectual correlates of reading. Reading was found to be significantly related to adaptive abilities. Findings highlighted the importance of appropriate and ongoing reading instruction for people with WS, including children and adults. The thesis provides evidence that an understanding of adaptive abilities in special populations such as WS is critical, not only for diagnostic purposes, but also to assist with planning appropriate educational, vocational and recreational opportunities and support for these individuals and their caregivers, to promote independence. A particularly strong message of the findings from this thesis is that adults with WS require ongoing efforts to ensure an enriched environment that leads to further opportunities for further education, social interaction, routine and personal growth.

Table of Contents

General Introduction

This thesis explores everyday living skills or adaptive functioning in Williams syndrome (WS). Adaptive functioning refers to an individual's performance on activities required for personal and social independence and includes communication skills, socialisation abilities, and daily living skills (Tasse et al., 2012). The overall purpose of this thesis is to investigate the adaptive profile of children and adults with WS and to examine demographic and environmental factors which may be related to their level of independence in daily life. Furthermore, the functional literacy skills of WS individuals and their relationship with adaptive functioning are investigated.

Williams syndrome is a neurodevelopmental disorder resulting from a microdeletion on chromosome 7 at *7q11.23*. The syndrome is associated with physical features (e.g., distinctive facial features, short stature), medical characteristics (e.g., cardiovascular abnormalities, hypercalcaemia), and intellectual and learning impairments, typically in the mild to moderate range (Martens, Wilson, & Reutens, 2008). Individuals with WS also have a distinctive personality and are described as very friendly, outgoing and empathic (Jones et al., 2000) but at the same time, they can experience psychological and behaviour impairments (see review by Martens et al., 2008). Individuals with WS are described as being more anxious and experience higher rates of generalised anxiety and specific fears and phobias (Dodd & Porter, 2009). Behavioural disturbance includes: being more distractible, inattentive and hyperactive; poor emotional regulation and low frustration tolerance; difficulty inhibiting inappropriate behaviours and being overly affectionate and more likely to experienced difficulties with peer relationship (Einfeld, Tonge, & Florio, 1997). It has been suggested that these social and behavioural characteristics may be the result of executive functioning impairment (e.g., Porter et al., 2012; Rhodes, Riby, Park, Fraser, & Campbell, 2010).

The WS cognitive profile has distinct peaks and valleys with weaknesses in nonverbal (e.g., visuo-spatial) abilities and relative strengths and weaknesses in certain aspects of verbal functioning. Relative verbal language strengths include auditory verbal short term memory and receptive vocabulary (Meyer-Lindenberg, Mervis, & Berman, 2006) while the comprehension and production of spatial language and pragmatic skills, are specific weaknesses (see review by Brock, 2007). Findings of verbal strengths and/or weaknesses depend on the nature of the task, choice of control group and aspect of verbal ability being assessed. There is evidence of considerable heterogeneity in the clinical, intellectual and cognitive functioning of individuals with WS (Jarrold, Baddeley, & Hewes, 1998; Porter & Coltheart, 2005).

The heterogeneity in WS makes it a particularly interesting syndrome to research, as it provides a unique opportunity to explore the relationship between intellectual and more specific cognitive levels and a wide range of variables of interest. WS is also associated with genetic variability, with many individuals with WS showing the standard ~1.6Mb deletion (involving approximately ~26 genes *at 7q11.23*), and others showing a larger 1.8Mb (28 gene) deletion (Antonell et al, 2010; Porter et al., 2012; Tassabehji, 2003). The current thesis takes the opportunity to explore this heterogeneity in WS.

The first chapter of this thesis consists of a literature review of relevant studies which have investigated adaptive functioning skills in WS. An electronic search of databases results in 15 published studies and 3 PhD dissertations being included in the review. Methodological limitations of the literature are considered, indicating the need for further research to consider changing adaptive profiles from childhood to adulthood. In addition, while some previous studies consider demographic variables, such as level of intellectual functioning and its relation to adaptive skills, it is concluded that environmental factors should also be considered.

The second chapter of the thesis is an empirical study following on from the conclusions of the review in Chapter 1. The main aim of the study in Chapter 2 was to investigate adaptive functioning and maladaptive (problem) behaviours in WS using an up-to-date assessment tool – the Vineland Adaptive Behavior Scale – Second Edition – (Vineland II; Sparrow, Cicchetti, & Balla, 2005), which has normative data that is appropriate across a wide age span, including children and adults. Thirty participants were assessed, all of whom had the standard ~1.6Mb deletion. Demographic variables of gender, chronological age and intellectual and cognitive functioning are considered in relation to adaptive functioning. For the first time, environmental factors, more specifically, family characteristics such as family relationships (e.g., cohesive, expressive), personal growth orientation (e.g., focus on intellectual, cultural or recreational activities) and family organisation levels - measured by The Family

Environment Scale - (FES; Moos & Moos, 1994) are considered. Detailed analyses are included both looking at group trends and considering individual variability at a case-by-case level.

The third chapter is comprised of an empirical study looking at reading skills in WS and how they may relate to adaptive functioning outcomes. Functional reading skills are the emphasis of this chapter, which refer to the minimum level of reading to enable an individual to perform everyday work and social activities and participate in the community. Thirty individuals with WS are included in the study and a comparison of 22 participants with a 1.6Mb deletion to eight who have a 1.8Mb deletion is undertaken. There is little consensus about what the level is at which an individual would be considered functionally literate. Guidance from Australian research indicated that the end of Year 5 of primary school be considered as the level at which individuals have reached a minimum level of competence in reading (Wheldall & Watkins, 2004), and was the level of reading performance used as the focus in this research. A cognitive neuropsychological approach is undertaken, whereby individuals were asked to read regular words, irregular words and nonwords, measured using the Castles and Coltheart Reading Test 2 (CC2; Castles et al., 2009). The CC2 is based on the Dual Route model of reading (Coltheart, Rastle, Perry, Langdon, & Ziegler, 2001), and allows for analysis of reading via a lexical versus nonlexical pathway. Reading abilities are considered at the group level, along with the relationship between reading and adaptive functioning. The reading ability of each individual is also profiled, resulting in subgroups of individuals based upon their performance on lexical and nonlexical reading. The relationship between intellectual functioning and cognitive skills shown to be related to reading ability in typically developing readers is also included.

The final chapter encompasses the overall contribution, conclusions and clinical implications of the thesis from a neuropsychological perspective.

References

Antonell, A., Del Campo, M., Magano, L.F., Kaufmann, L., Martinez de la Iglesia, J., Gallastegui, F., Flores, R., Schweigmann, U., Fauth, C., Kotzot, D., Perez-Jurado, L.A. (2010). Partial 7q11.23 deletions further implicate *GTFI* and *GTF2IRD1* as the main genes responsible for the Williams-Beuren syndrome neurocognitive profile. *Journal of Medical Genetics, 47,* 312-320.

Brock, J. (2007). Language abilities in Williams syndrome: A critical review. *Development and Psychopathology, 19,* 97-127.

Castles, A., Coltheart, M., Larsen, L., Jones, P., Saunders, S., & McArthur, G. (2009). Assessing the basic components of reading: A revision of the Castles and Coltheart test with new norms. *Australian Journal of Learning Difficulties, 14,* 67-88.

Coltheart, M., Rastle, K., Perry, C., Langdon, R., & Ziegler, J. (2001). DRC: A dual route cascaded model of visual word recognition and reading aloud. *Psychological Review, 108,* 204-256.

Dodd, H.F., & Porter, M.A. (2009). Psychopathology in Williams syndrome: The effect of individual differences across the life span. *Journal of Mental Health Research in Intellectual Disabilities, 2,* 89-109.

Einfeld, S.L., Tonge, B.J., & Florio, T. (1997). Behavioral and emotional disturbance in individuals with Williams syndrome. *American Journal on Mental Retardation, 102,* 45-53.

Jarrold, C., Baddeley, A.D., & Hewes, A.K. (1998). Verbal and non-verbal abilities in the Williams syndrome phenotype: Evidence for diverging developmental trajectories. *Journal of Child Psychology and Psychiatry, 39,* 511-523.

Jones, W., Bellugi, U., Lai, Z., Chiles, M., Reilly, J., Lincoln, A., & Adolphs, R. (2000). Hypersociability in Williams Syndrome. *Journal of Cognitive Neuroscience, 12,* 30-46.

Martens, M.A., Wilson, S.J., & Reutens, D.C. (2008). Research review: Williams syndrome: A critical review of the cognitive, behavioral, and neuroanatomical phenotype. *The Journal of Child Psychology and Psychiatry, 49,* 576-608.

Meyer-Lindenberg, A., Mervis, C.B., & Berman, K.F. (2006). Neural mechanisms in Williams syndrome: A unique window to genetic influences on cognition and behaviour. *Nature Reviews Neuroscience, 7,* 380-393.

Moos, R.H., & Moos, B.S. (1994). Family Environment Scale Manual (Third Edition). Palo Alto, CA: Consulting Psychologists Press.

Porter, M.A. & Coltheart, M., (2005). Cognitive heterogeneity in Williams syndrome. *Developmental Neuropsychology, 27,* 275-306.

Porter, M.A., Dobson-Stone, C., Kwok, J.B.J., Schofield, P.R., Beckett, W., & Tassabehji, M. (2012). A role for transcription factor GTF2IRD2 in executive function in Williams-Beuren syndrome. *PLoS ONE, 7,* e47457. DOI: 10.1371/journal.pone.0047457.

Rhodes, S.M., Riby, D.M., Park, J., Fraser, E., & Campbell, L.E. (2010). Executive neuropsychological functioning in individuals with Williams syndrome. *Neuropsychologia, 48,* 1216-1226.

Sparrow, S.S., Cicchetti, D.V., & Balla, D.A. (2005). Vineland Adaptive Behavior Scales (2nd Ed.). Survey Forms Manual. AGS Publishing, Circle Pines, MN: AGS Publishing.

Tassabehji, M. (2003). Williams-Beuren syndrome: A challenge for genotype-phenotype correlations. *Human Molecular Genetics, 12,* r229-r237.

Tasse, M.J., Schalock, R.L., Balboni, G., Bersani, H., Borthwick-Duffy, S.A., Spreat, S., Thissen, D., Widaman, K.F., & Zhang, D. (2012). The construct of adaptive behavior: Its conceptualization, measurement, and use in the field of intellectual disability. *American Journal on Intellectual and Developmental Disabilities, 117*, 291-303.

Wheldall, K., & Watkins, R. (2004). Literacy levels of male juvenile justice detainees. *Educational Review, 56,* 3-11.

The articles which comprise this thesis are co-authored by the supervisors of the doctoral candidate, Dr Melanie Porter and Dr Saskia Kohnen, Macquarie University

This work has not been submitted for a higher degree to

any other university or institution.

Signed by candidate (Gabrielle Brawn): __________________________________

Ethics Approval was obtained for this research

Protocol Number: HE27FEB2009-R063

Acknowledgements

I would like to thank all of my participants for taking part in this research as they have been an absolute joy to work with. Also, thanks to their wonderful families who gave up a lot of time for their family member to take part in our research. I would also like to sincerely thank my supervisors, Dr Melanie Porter and Dr Saskia Kohnen, for their assistance with all stages of this project. Thank you especially to Mel, for your unwavering support throughout difficult times of my candidature and for your invaluable knowledge and persistence. A special thank you to my immediate and extended family who have all been incredibly supportive of my studies.

Adaptive Functioning in Williams Syndrome: A Systematic Review

Gabrielle Brawn and Melanie Porter

Psychology Department, Macquarie University, Sydney, NSW, Australia

Correspondence should be sent to Melanie A. Porter, Psychology Department, Macquarie University, Sydney, Australia 2109. Phone: +61 2 9850 6768, Fax: +61 2 9850 8062, E-mail: melanie.porter@mq.edu.au

Abstract

Literature on the level of adaptive functioning and relative strengths and weaknesses in functioning of individuals with Williams syndrome (WS) was reviewed. The electronic databases PsycINFO, PubMed, Expanded Academic, Web of Science, Scopus and ProQuest were searched electronically for relevant articles and dissertations using the search terms "Williams syndrome" or "Williams-Beuren syndrome" combined with "adaptive function*", "adaptive behavio*", "independ*", and "autonomy". Selection criteria included English language articles, theses and book chapters, participants with a diagnosis of Williams syndrome and inclusion of a standardized assessment of adaptive functioning. Fifteen published articles and three Ph.D. dissertations met the selection criteria for inclusion in the review. Ten investigated adaptive functioning in children and adolescents aged up to 19 years of age, six investigated adaptive functioning in adults and two included participants across a wider age range and included both children and adults. Along with identifying methodological issues, the review addressed the following areas: overall level of adaptive functioning in WS, domain strengths and weaknesses, evidence of heterogenity, relationship to intellectual ability, changes with chronological age, relationship with maladaptive behaviour, gender differences, and results of other factors which may be related to adaptive functioning. This review highlights the need for further research into adaptive functioning in WS using appropriate and up-to-date assessment materials to further investigate the questions of heterogeneity and the potential influence of environmental factors on adaptive functioning in WS.

Introduction

Williams syndrome (WS), also referred to as Williams-Beuren syndrome, is a rare genetic disorder which results in specific physical, medical, behavioural and cognitive outcomes. WS is caused by a microdeletion typically of around 26 genes on the long arm of one copy of chromosome 7 at band q11.23. A diagnosis of WS can now be genetically confirmed by laboratory florescent in situ hybridization (FISH) analysis, which looks at one gene - the elastin gene (ELN) - located within the WS critical region and deleted in approximately 96% of cases (Lowery et al., 1995). The prevalence of WS is estimated to be about 1 in 7,500 live births (Strømme, Bjørnstad, & Ramstad, 2002).

Physical features of WS can include small stature, dysmorphic facial features, a hoarse voice, hyperacusis, and transient-neonatal hypercalcaemia. Medically, there can be connective tissue abnormalities, decreased motor coordination and balance, as well as reduced muscle tone and a high frequency of cardiovascular abnormalities, typically supravalvular aortic stenosis (SVAS) and peripheral pulmonary stenosis (PPS; Antonell et al., 2010; Williams, Barratt-Boyes, & Lowe, 1961). Behaviourally, individuals with WS are described as extremely friendly or “hypersociable” and they are noted for their willingness to eagerly engage in social interactions and to display affection, even with strangers (Bellugi, Lichtenberger, Jones, Lai, & St. George, 2000; Doyle, Bellugi, Korenberg, & Graham, 2004; Meyer-Lindenberg, Mervis, & Berman, 2006). At the same time, individuals with WS can experience generalized anxieties, fears and specific phobias, and have difficulties with attention, and can be distractible and hyperactive (see review by Martens, Wilson, & Reutens, 2008).

Infants and young children with WS typically present with developmental delay (Mervis & Klein-Tasman, 2000). Intellectual and learning disabilities are also reported in most individuals with WS, with overall functioning typically at the level of a mild to moderate impairment (Martens et al., 2008; Porter & Coltheart, 2005), although the intellectual disability level has been found to range widely from severe intellectual disability to the average to low-average range (Mervis & John, 2010). Likewise literacy skills range widely from an inability to read to average for age. Moreover, there have been reports of specific strengths and weaknesses in the cognitive profile of WS, characterized by relative strengths in certain verbal abilities including: receptive vocabulary; grammatical abilities; verbal short-term memory; and (concrete) nonverbal reasoning, but with considerable weaknesses in: visuospatial constructional abilities; relation/conceptual language (spatial, dimensional, and temporal concepts) and pragmatics (Mervis & Klein-Tasman, 2000; Mervis & John, 2010). Significant individual variability in cognitive strengths and weaknesses has been demonstrated (Jarrold , Baddeley, & Hewes, 1998; Pezzini, Vicari, Voltera, Milani & Ossella, 1999; Porter & Coltheart, 2005; Tassabehji et al., 1999).

While the intellectual functioning and cognitive profile of individuals with WS has received a great deal of attention in the literature to-date, investigations into how well an individual with WS functions in their day-to-day environment (adaptive functioning) has received less focus. Adaptive functioning or adaptive behaviour is the collection of conceptual, social, and practical skills that individuals must learn in order to enable them to function in their everyday lives and which, ultimately, enable them to live independently (Venn, 2007). A person who has deficits in their adaptive functioning can experience difficulties in meeting the demands of various environments and situations and may be dependent for certain daily living skills (Harrison & Boney, 2002). For example, children and adults who have deficits in adaptive functioning may experience difficulties with important life activities such as interacting with peers, taking care of their personal needs, learning new skills, and general functioning in natural environments such as home, school, work and the community (Harrison & Boney, 2002).

Appropriate assessment of adaptive functioning relies upon understanding the underlying construct to be measured; however, there has been criticism and debate in the literature regarding the lack of a consensual theoretical definition and the structure of the construct (Dixon, 2007; Thompson, McGrew, & Bruininks, 2002). Investigations using factor analysis suggest that adaptive functioning has a multifactorial or multidimensional structure (Harries et al., 2005) and five factors have been proposed (personal independence, responsibility, cognitive/academic performance, physical/developmental competencies, and vocational/community skills), along with maladaptive factors (Schalock, 2004). An important characteristic of adaptive functioning is that adaptive skills are age-related (Sparrow, Cicchetti, & Balla, 2005) and, as such, adaptive functioning is expected to develop and increase as an individual becomes older and as the demands of the environment become more complex (Harrison

& Boney, 2002). Also, adequate adaptive functioning depends on the ability of the person to match skills to the current environment and to change their behaviour depending on the current demands (Harrison & Boney, 2002). While an individual may have the necessary ability to perform a task, their adaptive functioning would be considered inadequate if they failed to perform the task when required (Sparrow et al., 2005). To better convey these concepts, more descriptive terms such as everyday competence or typical competence behaviour have been proposed (Thompson et al., 2002).

Standardised tests (scales) are employed in assessing an individual's adaptive functioning and, due to the difficulty in observing the typical daily living functioning in real-life settings, rely on indirect measurement by informant report, typically a parent or primary caregiver (Venn, 2007). Assessment of adaptive functioning aims to measure an individual's typical performance on day-to-day tasks and is different to the assessment of intellectual ability, which endeavours to measure an individual's cognitive performance (Hogan, 2003). For example, a test of intellectual functioning may test the limits of an individual's vocabulary, while an adaptive behaviour scale would aim to look at the words the individual typically uses on a daily basis (Hogan, 2003).

Assessment of adaptive functioning is utilised in the definition and diagnosis of intellectual disability (ID). For example, the American Association on Intellectual and Developmental Disabilities (AAIDD) 2010 definition of intellectual disability refers to significant limitations in both intellectual functioning and adaptive behaviour in order to make a formal diagnosis of intellectual disability. The AAIDD definition of adaptive behaviour is described as comprising three dimensions: conceptual skills (e.g., communication, language, academic skills) social skills (e.g., social interaction and social problem solving), and practical skills (e.g., daily living skills, personal care, travel, safety). Similarly, the American Psychiatric Association's (APA) definition of intellectual disability (using the outdated term 'Mental Retardation') in the Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition Text Revision (DSM-IV TR; APA, 2000) refers to significant deficits or impairments in adaptive functioning in at least two of the following skill areas: communication, self-care, home living, social/ interpersonal skills, use of community resources, self-direction, functional academic skills, work, leisure, health, and safety along with significantly sub-average intellectual functioning (DSM-IV TR, 2000). With the release of the fifth revision of the DSM (DSM 5; APA, 2013), not only has the terminology been revised to Intellectual Disability (Intellectual Developmental Disorder), but also a greater emphasis has been placed on deficits in adaptive functioning in the diagnostic criteria. While deficits in both intellectual (IQ) and adaptive functioning continue to be required for diagnosis, the level of impairment (mild, moderate, severe or profound intellectual disability) is determined by an individual's level of adaptive functioning, not IQ scores, as such adaptive functioning will determine the level of supports the individual will require (APA, 2013).

While both intellectual and adaptive functioning play a role in the diagnosis of ID, there has been little research and consensus into the important question about the relationship between standardized measures of adaptive functioning and intellectual functioning (IQ) and while there is evidence of a relationship between the two, the exact relationship is unknown (Sattler, 2008). Sparrow et al. (2005) stated that a relationship would be expected between adaptive domains, which rely on academic ability and intelligence scales, as they both measure academic skills. An early review of studies by Harrison (1987) found that correlations between adaptive functioning and IQ varied widely from .03 to .91, with the majority indicating a moderate relationship. In a more recent research review, McGrew (2012) conducted an informal research synthesis and concluded that a reasonable estimate of the adaptive functioning/IQ correlation is approximately .50, with most correlations ranging from .40 to .65, which is consistent with Harrison's finding of moderate correlations. McGrew concludes that these findings indicate that "adaptive behavior and intelligence are statistically related constructs, but they are still independent". Harrison noted that, while measurement of adaptive behaviour and IQ has similar purposes, there are several differences between them. Specifically: IQ scales assess thought processes, while adaptive functioning scales measure everyday behaviours; IQ aims to measure maximal performance, while adaptive functioning measures an individual's typical performance; and there is a presumption of stability in IQ scores, while adaptive functioning is presumed to be modifiable (Harrison, 1987). Correlations between adaptive functioning and IQ tend to be higher for individuals within the severe range of intellectual disability compared to those within the mild range of disability (Sattler, 2008).

Assessment of Adaptive Functioning

Assessment of the adaptive functioning skills of individuals with WS can provide information about the impact of WS and its unique cognitive and behavioural phenotype on real-world functioning (Mervis, Klein-Tasman, & Mastin, 2001). Rather than just undertake a syndrome specific view to adaptive abilities, it is important to also consider within-syndrome heterogeneity in the area of adaptive functioning. Therefore, while the absolute level of functioning is important, relative strengths and weaknesses and the relationship between adaptive functioning and IQ and other cognitive abilities should also be considered. This information can, in turn, provide important information for families, special educators and employers in identifying the supports needs of WS individuals and to develop appropriate intervention programs. For example, information can be used to help develop individual education programs (IEPs) for students and suitable work placements for adults. As successful employment and independent living are dependent upon adaptive skills (Mervis & Morris, 2007) appropriate services and supports can assist individuals with WS to live productive and fulfilling lives (Thompson et al., 2002). Therefore, systematic research is important to provide useful information, which can result in better outcomes in areas of daily independence, education, and learning (Thompson et al., 2002). Adaptive functioning is also an area in which caregivers of individuals with WS have an opportunity to make an impact on skill development. However, parental attitudes may affect the emphasis which is placed on acquisition of self-help skills, for example, how much responsibility they expect their child to undertake for household chores (Mervis & Morris, 2007).

The purpose of this review paper was to systematically examine the literature to investigate the level of adaptive functioning of children, adolescents and adults with Williams syndrome and individual patterns of strengths and weaknesses. The relationships between adaptive functioning with both intellectual functioning and chronological age will be examined, along with gender differences.

Method

A search was conducted of the databases PsycINFO, PubMed, Expanded Academic, Web of Science and Scopus. Search terms included: (a) “Williams syndrome” or “Williams-Beuren syndrome” combined with the Boolean operator “AND” with the terms (b) “adaptive function*”, “adaptive behavio*”, “independ*”, and "autonomy". The reference lists of identified articles were also reviewed for relevant papers and book chapters. All articles up to March 2013 were included with no limit on the period covered. In an attempt to reduce publication bias, a search of the ProQuest database, which provides access to full text dissertations and theses, was undertaken. The following selection criteria were applied: (1) English language articles/theses/book chapters, (2) participants formally diagnosed with Williams syndrome, (3) inclusion of a standardised assessment of adaptive functioning. As discussed earlier, adaptive functioning is best conceptualised as being multi-dimensional, and, therefore, only papers that included assessment of functioning across a range of areas or domains (such as daily living skills, socialisation, and communication skills) were included. Articles that examined only one facet of adaptive functioning (for example, social functioning only) were excluded. Genetic confirmation of Williams syndrome via fluorescent in situ hybridization (FISH) techniques has only recently become a common feature of published articles. As there were only a small number of articles that have investigated adaptive functioning in Williams syndrome, studies that used clinical diagnosis based on physical (e.g., short stature) and medical (e.g., supravalvular aortic stenosis) features were also included.

After applying these criteria, 15 published studies and 3 Ph.D. dissertations were identified and are summarized in Table 2. Ten studies had investigated adaptive functioning in children and adolescents (oldest individual was aged 19 years); six studies investigated adaptive functioning in adults (all individuals over 18 years); and two studies investigated adaptive functioning across the age range from childhood through to adulthood. Three studies were excluded over and above the 15. A study by Edgin, Pennington, and Mervis (2010) investigated memory patterns (immediate, working and associative memory) in 28 individuals with WS aged from 12 years through to 26 years. They also included a measure of adaptive behaviour (Scales of Independent Behaviour – Revised, SIB-R) in an attempt to ascertain which memory types are related to variation in adaptive abilities. While an overall Broad Independence standard score was provided, the scores for the four clusters which contributed to this overall score were not detailed and as such, this study was not included in the systematic review. One published article (Crawford, Edelson, Skwerer, & Tager-Flusberg, 2008) and one dissertation (Philofsky, 2006) were excluded as they had measured only one aspect of adaptive functioning (Socialisation).

Critical Evaluation on Studies of Adaptive Functioning in WS

Methodological quality was not an eligibility criterion, but was assessed for each paper using the following criteria: adequate sample size defined as at least 20 participants; diagnosis of WS confirmed genetically with FISH test; the use of a standardized, reliable and valid measure of adaptive functioning; means and standard deviations provided for overall/composite measure of adaptive functioning; means and standard deviations of domains of adaptive functioning provided; use of appropriate statistical methods, for example corrections to minimise Type 1 errors. The criteria included items raised by Martens et al.'s 2008 research review. For example, the sample size of at least 20 was selected following Martens et al.'s comments that the median sample size of past studies has ranged from 6 to 17 participants, and these smaller sample sizes had limited generalizability of findings.

Methodological issues are summarized in Table 1. Group sample size was small in several studies (Cherniske et al., 2004; Di Nuovo & Buono, 2011; Fisch et al., 2010; Gosch & Pankau, 1994; Greer, Brown, Shashidhar Pai, Choudry, & Klein, 1997; Plissart, Borghgraef, Volcke, Van den Berghe, & Fryns, 1994) due to the practical limitations of studying a rare syndrome. Genetic confirmation of WS via a positive FISH analysis has only been available since the 1990s, so only more recent papers have employed participants who have all had genetic confirmation of WS diagnosis (Dimitropoulos, Ho, Klaiman, Koenig, & Schultz, 2009; Fisch et al., 2007; Fisch et al., 2010; Fu, 2012; Howlin, Elison, Udwin, & Stinton, 2010 [cross-sectional study only]; John & Mervis, 2010; Mervis & John, 2010; Rowe, 2007). Phillips (2008, p. 46) refers to the fact that all participants had "confirmed diagnoses of Williams syndrome" which is assumed to mean genetic confirmation; however this is not clear. Martens et al. (2008, p. 599) commented that "the scientific rigor of WS research would be enhanced if future studies continued to make every effort to employ participants who have a genetic confirmation of WS". While genetic confirmation via a positive FISH analysis has been included in more recent studies, there is still the possibility of heterogeneity within the WS sample due to variations in the size of an individual's deletion (Porter et al., 2012). Porter et al. suggest, for example, that the prevalence of the atypical ~1.8Mb deletion may be higher than previous estimates of 5% and as high as 18%, at least in their Australian cohort, and may have implications for cognitive and behavioural functions. Only one other study to-date (Fu, 2012) has excluded individuals with an atypical deletion, stating “WS individuals without the typical deletion were excluded from this study, since they presented a slightly different picture from individuals with WS with typical deletions” (Fu, 2012, p. 26). However, it is unclear whether a full genotype screen was conducted.

Reliability and validity of the measure of adaptive functions each study employed was considered appropriate for the majority of studies[1]. Two studies used German forms of questionnaires making it difficult for the reviewer to assess their appropriateness (due to the language barrier). Gosch and Pankau (1994) used a German short form of the Vineland Social Maturity Scale, and the authors provided statistical information regarding its reliability (inter-rater reliability, internal consistency, re-test reliability). Plissart et al. (1994) used two German standardized questionnaires and the authors stated that the reliability and validity was “sufficient” but they did not provide further details. Not all studies provided an overall score for adaptive functioning (Davies, Howlin, & Udwin, 1997; Fisch et al., 2010; Howlin, Davies, & Udwin, 1998) or standard deviations or ranges of scores (Cherniske et al., 2004). Most papers presented domain standard scores, although three papers presented data in graphical form only (Fisch et al., 2007; Fisch et al., 2010; Plissart et al., 1994), making it difficult to compare results to other studies. Statistical methodology was limited by failure to apply statistical analyses to compare domain scores (Cherniske et al., 2004; Davies et al., 1997; Fisch et al., 2010; Phillips, 2008; Plissart et al., 1994; Rowe, 2007) or by failing to state whether they had adequately corrected for multiple statistical comparisons (Greer et al., 1997; Howlin et al., 1998; Mervis & John, 2010) to control for Type 1 error. However, it should be noted that with small sample sizes it may be considered preferable to make a Type 1 error by not making adjustments for multiple comparisons, rather than make a Type II error (see Rothman, 1990). Only one study included effect size statistics making it transparent as to whether statistical power was an issue in the study (John & Mervis, 2010).

[1] The measures used had acceptable psychometric properties (reliability and validity), uniform procedures for administration and scoring and normative data for comparison of results. Information on the psychometric properties for each measure can be found in the relevant technical manual.

Table 1

Methodological quality of included studies

Study	*Adequate sample size*	*Diagnosis genetically confirmed*	*Reliable & valid measure of adaptive functioning*	*Means (sd) of overall adaptive functioning provided*	*Means (sd) of domains provided*	*Appropriate statistical methods*
Gosch & Pankau (1994)	No	No	Yes	Yes	No	Yes[g]
Plissart et al. (1994)	No	No	No[a]	Yes	No[b]	No[b]
Davies et al. (1997)*	Yes	No	Yes	No	Yes	No[b]
Greer et al. (1997)	No	No[e]	Yes	Yes	Yes	No[c,g]
Howlin et al. (1998)*	Yes	No	Yes	No	Yes	No[c]
Mervis et al. (2001)	Yes	No[e]	Yes	Yes	Yes	Yes[g]
Cherniske et al. (2004)	No[d]	No[e]	Yes	Yes[f]	Yes[f]	No[c]
Fisch et al. (2007)	Yes	Yes	Yes	Yes	No[b]	Yes
Rowe (2007)	Yes	Yes	Yes	Yes	Yes	Yes[g]
Phillips (2008)	Yes	No[e]	Yes	Yes	Yes	Yes[g,]
Dimitropoulos et al. (2009)	Yes	Yes	Yes	Yes	Yes	Yes
Fisch et al. (2010)†	No	Yes	Yes	No[b]	No[b]	No[c]
Howlin et al.* (2010) Cross-sectional[a]	Yes	Yes	Yes	Yes	Yes	Yes[g]
Howlin et al.* (2010) Longitudinal[a]	Yes	No	Yes	Yes	Yes	Yes[g]
John & Mervis (2010)	Yes	Yes	Yes	Yes	Yes	Yes
Mervis & John (2010)	Yes	Yes	Yes	Yes	Yes	No[c]
Di Nuovo & Buono (2011)	No	Yes	Yes	No	No[b]	Yes[g]
Fu (2012)	Yes	Yes	Yes	Yes	Yes	No[c]

Note. * Related studies involving the same participants, †Longitudinal follow-up of Fisch et al. (2007); Howlin et al. (2010) also reported in Elison, Stinton, & Howlin (2010).[a]Reliability and validity of scale described as “sufficient” but no details provided; [b]Results presented in graphical form ; [c] No details of statistical analysis to compare domains and subdomains reported; or multiple statistical tests performed with inadequate or no mention of correction of for Type 1 error; [d] Data for VABS collected on 18 participants; [e]Genetic confirmation for some but not all participants or criteria for diagnosis not explicitly stated; [f]No standard deviations provided, nor range of scores; [g]No measure of effect size provided.

Results

A descriptive synthesis of the data provided by the studies was undertaken. As noted by Mervis et al. (2001), age equivalency (AE) scores typically violate many assumptions of statistical analyses. Age equivalency scores are based on an ordinal scale and the scale units are unequal, making comparisons difficult (Sattler, 2008). Therefore, only results of standard scores will be utilised in this review. As all studies provided overall adaptive and domain level standard scores, no study was excluded on this basis. The aim of several papers (Dimitropoulos et al., 2009; Di Nuovo & Buono, 2011; Fisch et al., 2007; Fisch et al., 2010) was to investigate between-group differences, comparing adaptive functioning in WS to groups with intellectual disability or other developmental disorders (for example, Down Syndrome). As the focus of this paper was within-group adaptive behaviour patterns in William syndrome, only the data related to WS was extracted and considered in this review. Between-group studies are important and early studies of genetic conditions focused on this to provide evidence that particular genetic conditions differed in their behaviour (Dykens & Hoddap, 2007). However, as acknowledged by Dykens and Hoddap (2007), while between-group differences are well established, research is focusing on within-syndrome approaches to provide information about their characteristic behaviour, individual differences and possible causes of variability such as genetics.

The most commonly used adaptive behaviour scale across all studies was the Vineland Adaptive Behavior Scales (VABS: Sparrow, Balla, & Cicchetti, 1984) which was utilized in 12 out of the 18 studies (see Table 2). One dissertation (Rowe, 2007) used the Vineland Adaptive Behavior Scales, Second Edition (Vineland II; Sparrow et al., 2005) across a narrow age range (3 to 5 years). The VABS measures adaptive behaviour in the domain areas of Communication, Daily Living Skills, Socialisation, and Motor Skills (for individuals under 7 years of age) and the domains combine to provide an overall measure of adaptive functioning (Adaptive Behavior Composite or ABC). Normative data is available for ages 0 to 18 years, which does not cover the age of participants in the six studies involving adults (ages included in these studies ranged from 19 to 55 years) in which the VABS was used. The Vineland II maintains the same four domain structure of the VABS, but new items have been added to improve measurement. The norms have been extended to cover ages from 0 to 90 years and standardized scores (v-scale scores) are available for each subdomain, allowing statistical analysis of functioning at the subdomain, domain and overall level. Both the VABS and Vineland II have an optional Maladaptive Behavior domain, and the Vineland II allows separate measurement of Internalising and Externalising behaviours. The Scales of Independent Behavior – Revised (SIB-R; Bruininks, Woodcock, Weatherman, & Hill, 1996) was used in four studies. The SIB-R assesses competency in Motor Skills, Social Interaction and Communication Skills, Personal Living Skills, and Community Living Skills and includes an overall measure of adaptive functioning (Broad Independence) and an optional Problem Behavior scale. The SIB-R covers ages from infancy to 80 years and therefore provided appropriate normative data for all participants assessed. Both the VABS (and Vineland II) and SIB-R have been standardized on the same scale, with a mean of 100 and standard deviation 15. Two early studies (Gosch & Pankau, 1994; Plissart et al., 1994) used German adaptations of scales (the Vineland Social Maturity Scale and Cain-Levine competency scale respectively).

Table 2

Summary of Results from Included Studies

Study	Sample size [age range in years]	Measures	Overall Adaptive Functioning and Domains Means (SD) [range]	Findings
Gosch & Pankau (1994)	WS = 19 [age 4-10] ID = 19 [age 4-10]	VSMS Checklist; Columbia Mental Maturity Scale; CBCL	VSMS mean = 18.22 (5.35)	Nonverbal IQ = 79 (13.0) WS children significantly less well-adjusted on social-emotional adjustment and personal independence compared to control group
Plissart, et al. (1994)	WS = 11 [age 17-66]	SRZ(Autonomy); SGZ (Behavior); McCarthy Scales	SRZ mean = 5.83 scale mean score = 6.33 (1.33)	Highest score on language and social orientation Lowest scores on personal care/daily living skills
Davies et al. (1997)*	WS = 70 [age 19-39]	VABS; WAIS-R; BPVS; EOWVT; WORD; RPCM	ABC not provided Comm: 34.17 (19.26) DLS: 36.53 (19.42) Social: 44.61 (17.65)	FSIQ = 62.00 (6.76) [Range: 46-84] Correlations between ABC with FSIQ: (Comm = .70, DLS = .57, Social,= .38)
Greer et al. (1997)	WS = 15 [age 4-18]	VABS; SBFE; CBCL	ABC = 54.13 (17.08) [24 – 83] Comm: 62.47 (20.05) [<20 – 89] DLS: 50.60 (20.47) [<20 – 84] Social: 63.13 (16.12) [37-90]	FSIQ= 62.33 (11.82) [Range: 43-80] No difference between ABC and FSIQ

				Comm > DLS (p < .001) Social > DLS (p < .001) No gender differences in adaptive behaviour.
Howlin et al. (1998)*	WS = 62 [age 19-39]	VABS; BPVS; EOWVT; WAIS-R; WISC-R; WORD	ABC not provided Comm: 29.53 (11.07) [19-81] DLS: 33.92 (17.32) [19-89] Social: 42.68 (14.85) [19-88]	FSIQ = 60.85 (5.94) Comm < DLS (*p* < .009) DLS < Social (*p* < .001) Social > Comm (*p* < .001)
Mervis et al. (2001)	WS = 41 [age 4-8]	VABS; DAS	ABC = 62.98 (9.33) [41 -90] Comm: 71.05 (12.12) DLS: 60.20 (10.09) Social: 78.70 (13.71) Motor (n = 18): 56.89 (8.28)	FSIQ (GCA) = 59.32 (11.84) [Range: 26 – 78] No difference between ABC and GCA Social > Comm and DLS; Comm > DLS Correlations between GCA with: (ABC = .41; Comm = .42; DLS = .34; Motor skills = .59). Correlation between ABC and age = .03
Cherniske et al. (2004)	WS = 20 [age 30-51]	VABS; WAIS-III, KBIT, DMR; ADIS; SADS	ABC: 55 Comm: 46 DLS: 61 Social: 65	Mean FSIQ = 68
Fisch et al. (2007)	WS = 34 [3-16]	VABS; SBFE; DBC-P	ABC: 57.3 (12.9) Social > DLS	Mean IQ = 52.1 (10.7) IQ < ABC (p < .001)

	FRAXA = 44 [3-14] NF1 = 30 [4-14]		Comm > DLS	Correlation between ABC with CA: -0.65. Little change in maladaptive behaviour with age.
Rowe (2007) Ph.D Dissertation	WS = 31 [3-5] DS = 34 [3-5]	Vineland II; Mullen Scales of Early Learning; IAMM; DMQ	ABC = 69.5 (6.5) [57-81] Comm: 76.2 (7.0) [57-89] DLS: 69.8 (9.2) [55-93] Social: 75.2 (8.3) [61-90] Motor: 69.9 (6.4) [59-84]	Mullen Scales = 59.8 (7.7) [Range = 49-78] Correlation ABC with Mullens: r = .55 Parent ratings of competence on DMQ significantly correlated with Vineland II (*r* = .47) for both WS and DS. Weak, non-significant correlations between task persistence on the IAMM and Vineland II scores for both WS and DS
Phillips (2008) Ph.D Dissertation	WS = 37 [8-15]	SIB-R; KBIT-II; D-KEFS; DCCS; TAPS; BRIEF; CBCL; ERC	SIB-R: 44.78 (16.12) Social/Comm: 68.78 (12.45) Pers Living: 55.05 (15.40) Home/Comm: 43.89 (20.06) Motor Skills: 49.05 (16.25)	FSIQ = 66.86 (11.55) [Range: 45-94] Negative correlations between Emotional Regulation and (SIB-R = -.51, Motor Skills = -.44, Personal Living = -.60)
Dimitropoulos et al. (2009)	WS = 20 [4-19] PWS = 31	VABS; WAIS-R; WISC-III; WPPSI-R; DBC-P	ABC: 53.0 (16.48) Comm: 60.47 (19.0) DLS: 45.9 (22.0)	FSIQ = 56.89 (10.94) FSIQ correlated with: (ABC: 0.56; Comm: 0.62; DLS:

	Autism = 61		Social: 67.57(14.1)	0.52) Correlation between Comm and age = -0.59
Fisch et al. (2010)†	WS = 18 FRAXA = 37 NF1 = 10	VABS; SBFE	Not provided	Negligible decrease in VABS from Time 1 to Time 2 (-0.22±6.93) No gender differences
Howlin et al. (2010)*	Cross-sectional: WS = 92 [19-55] Divided into 3 age groups: 1) 19 – 29 (n=44) 2) 30 – 39 (n=31) 3) 40 – 55 (n=17)	VABS; WAIS-III; BPVS, EOWPVT	Group 1: ABC: 35.1 (12.4) [19-65] Comm: 30.6 (15.0) [19-84] DLS: 37.2 (18.5) [19-83] Social: 46.8 (14.1) [19-90] Maladaptive raw score: 13.07 (8.01) [2-37] Group 2: ABC: 45.1 (18.5) [19-97] Comm: 37.4 (21.3) [19-92] DLS: 55.3 (26.6) [19-110] Social: 53.2 (18.8) [19-109] Maladaptive raw score: 7.74 (4.76) [2-23] Group 3: ABC: 42.9 (17.9) [20-95] Comm: 36.2 (19.9) [19-97] DLS: 51.35 (23.1) [19-95] Social: 51.50 (18.7) [27-98]	FSIQ Grp 1: 55.7 (6.8) [45-74] FSIQ Grp 2: 57.8 (7.3) [45-72] FSIQ Grp 3: 57.00 (8.1) [48-80] Grp 1: DLS < Social DLS Grp 2 > Grp 1 For the whole group: Correlations between FSIQ with (ABC = .63,Comm = .62, DLS = .59, Social,= .56) Correlations between PIQ and ABC = .57 VIQ and ABC = .59

			Maladaptive raw score: 12.19 (6.58) [1 -25]	
	Longitudinal study Time 1 (T1): WS = 47 [19-38] Time 2 (T2) WS = 47 [25-49]	VABS T1: WAIS-R T2: WAIS-III	T1: ABC: 33.3 (10.2) [19-53] Comm: 31.3 (13.4) [19-81] DLS: 32.98 (13.74) Social: 41.3 (12.7) [19-68] T2: ABC (T2) = 44.7 (16.1) [19-85] Comm: 33.6 (15.8) [19-89] DLS: 56.53 (26.33) [19-113] Social: 55.2 (19.6) [19-98]	Maladaptive decreases from T1 to T2
Elison et al. (2010)*	Cross-sectional: WS = 92 [19-55] Longitudinal: Time 1: WS = 49 [18-37] Time 2: WS = 49 [30-49]	VABS; WAIS-III; BPVS; EOWVT		Results reported in Howlin et al. (2010)
John & Mervis (2010)	WS = 78 [4-10]	SIB-R; SSP; KBIT-II; PPVT-III; BRIEF; CBQ; CPRS-R	SIB-R: 53.21 (19.34) [0 – 93] High Sensory Impaired Group: Social/Comm: 61.87 (18.78) Pers Living: 45.55 (16.78) Comm Living: 48.06 (18.88) Motor : 51.63 (16.11)	FSIQ = 76.76 (15.40) [Range: 40 -111] Statistically significant difference between High vs Low sensory impairment groups on each subscale of VABS with high sensory

			Low Sensory Impaired Group: Social/Comm: 73.89 (17.58) Pers Living: 64.47 (17.27) Comm Living: 62.56 (16.78) Motor: 63.86 (15.36)	impairment group having lower functioning.
Mervis & John (2010) also reported in Mervis & Morris (2007)	WS = 122 [4-17]	SIB-R; DAS	SIB-R: 55.11 (15.45) [24-95] Social/Comm: 73.16 (14.72) [30-110] Pers Living: 61.22 (14.53) [24-98] Comm Living: 57.35 (17.20) [24-96] Motor: 57.82 (15.13) [24-88]	GCA = 64.56 (12.33) [Range: 31-96] Mean SIB-R < mean GCA Correlation between SIB-R and age = -.31
Di Nuovo & Buono (2011)	WS = 12 [7-30] DS = 109 [4-39] AS = 16 [5-33] PWS = 18 [3-32] Fragile-X = 26 [4-36]	VABS (Italian version); Wechsler Intelligence Scale or other age appropriate scale (e.g., Griffiths Mental Development Scale)	ABC not provided Maladaptive: 10.37 (2.11)	Signif negative correlations between Maladaptive Behaviour and Comm (-.48), DLS (-.41)), Social (-.53) domains None of the correlations between VABS domains and IQ were significant No correlations between VABS and age
Fu (2012) Ph.D Dissertation	WS = 100 [12-52]	SIB-R; WISC-R; WISC-III; WAIS-	SIB-R = 48.47 Adjusted Means:	FSIQ = 64 (10) Verbal short-form IQ = 76.10

	DD = 25 [12-46]	R; WAIS-III; VMI; MPQ; SISQ, CBCL	Social/Comm: 67.52 Comm Living: 50.38 Pers Living: 58.67 Motor: 52.74	(10.14) Estimated verbal IQ correlated with (SIB-R,= .297; Social/Comm = .37; Pers Living,= .205; Comm Living = .392) Significant negative correlation between Social/Comm and age (-.208). Visual-motor functioning contributed significantly to non-social domains (No gender differences

Note. * Related studies using the same participants; †Longitudinal follow-up of Fisch et al. (2007).

Sample Characteristics: AS = Angelman Syndrome; DD = Developmental disabilities; DS = Down syndrome; FRAXA = Fragile X mutation; ID = Intellectual disability; NF1 = Neurofibromatosis type 1; PWS = Prader-Willi syndrome; WS = Williams-Beuron syndrome.

Scores: ABC = Adaptive Behavior Composite of Vineland; CA = chronological age; Comm = Communication; Comm Living = Community Living Skills; DLS = Daily Living Skills; FSIQ = Full scale Intellectual Quotient; Motor = Motor Skills; Pers Living = Personal Living Skills; SIB-R = Broad Independence Score; Social = Socialisation; Social/Comm = Social Interaction and Communication Skills. Unless otherwise stated, scores in the 'Overall Adaptive Functioning and Domains' and 'Findings' columns are based on standardized score with Mean = 100 and

Standard Deviation = 15. Higher score on overall measures of adaptive functioning and domains indicates better functioning. Maladaptive Scales measure problem behaviours, with a higher score indicating more problem behaviours.

Abbreviations for Table 2:

ADIS	Anxiety Disorder Interview Scale
BRIEF	Behavior Rating Inventory of Executive Functions;
BPVS	British Picture Vocabulary Scale
CBCL	Child Behavior Checklist
CBQ	Children's Behavior Questionnaire
CPRS-R	Conners' Parent Rating Scale – Revised
DAS	Differential Ability Scale
DBC-P	Developmental Behavior Checklist – Primary Carer Version, Second Edition
DCCS	Dimensional Change Card Sort
D-KEFS	Delis-Kaplan Executive Function System
DMQ	Dimensions of Mastery Questionnaire
DMR	Dementia Questionnaire for Persons with Mental Retardation
EOWVT	Expressive One-Word Expressive Vocabulary Test
ERC	Emotion Regulation Checklist

IAMM	Individualized Assessment of Mastery Motivation
KBIT	Kaufman Brief Intelligence Test
KBIT-II	Kaufman Brief Intelligence Test – 2nd Edition
McCarthy Scales	McCarthy Scales of Children's Abilities
MPQ	Multidimensional Personality Questionnaire
PPVT-III	Peabody Picture Vocabulary Test-III
RPCM	Raven's Progressive Coloured Matrices
SADS	Schedule for Affective Disorders and Schizophrenia
SIB-R	Scales of Independent Behavior – Revised
SBFE	Stanford-Binet Intelligence Scale – Fourth Edition
SGZ	Storend Gedragsschaal (measure of undesirable behaviour)
SISQ	Salk Institute Sociability Questionnaire
SRZ	Sociale Redzaamheidsschaal (an adaptation of the Cain-Levine Social Competency Scale)

SSP	Short Sensory Profile
TAPS	Test of Auditory-Perceptual Skills
VABS	Vineland Adaptive Behavior Scale
Vineland II	Vineland Adaptive Behavior Scales, Second Edition
VMI	Berry-Buktenica Developmental Test of Visual-Motor Integration, 4th Edition
VSMS	Vineland Social Maturity Scale
WAIS-R	Wechsler Adult Intelligence Scale – Revised
WAIS-III	Wechsler Adult Intelligence Scale – Third Edition
WASI	Wechsler Abbreviated Scale of Intelligence
WISC-R	Wechsler Intelligence Scale for Children – Revised
WISC-III	Wechsler Intelligence Scale for Children – Third Edition
WORD	Wechsler Objective Reading Dimensions
WPPSI-R	Wechsler Preschool and Primary Scale of Intelligence – Revised.

Overall level of adaptive functioning

As can be seen in Table 2, the overall level of adaptive functioning of children, adolescents and adults with WS is typically at a 'Low' level, being more than two standard deviations below the mean (i.e., < 70) on standardized assessment scales. The range of scores obtained across studies indicates a wide range of functioning for all age groups. While some individuals were found to be functioning four to five standard deviations below the population mean, which would be considered Profoundly or Severely impaired (Greer et al., 1997; Howlin et al., 1998; Howlin et al., 2010; Mervis et al., 2001; John & Mervis, 2010; Mervis & John, 2010), there is evidence that some individuals were functioning at a Moderately Low to Adequate (Average) level for their age (Greer et al, 1997; Howlin et al, 1998; Howlin et al., 2010; John & Mervis, 2010; Mervis et al., 2001; Mervis & John, 2010; Rowe, 2007).

Ten studies assessed children under 18 years and six studies provided information for adults (18 years plus), while two studies included participants aged across a wider age range (both children and adults). For children and adolescents with WS, the mean adaptive functioning score has been found to range from 44.78 (Phillips, 2008) to 69.5 (Rowe, 2007), while for adults with WS the reported range is from 33.3 (Howlin et al., 2010) to 55 (Cherniske et al., 2004). Information about adaptive functioning in adults is somewhat limited by the fact that four out of the six studies were related and involved many of the same participants (Davies et al., 1997; Elison et al., 2010; Howlin et al., 1998; Howlin et al., 2010).

Domain Strengths and Weaknesses

For children and adolescents, skills related to Socialisation and Communication skills were found to be higher than Personal/Daily Living Skills (Dimitropoulos et al., 2009; Fisch et al., 2007; Greer et al., 1997; John & Mervis, 2010; Mervis et al., 2001; Phillips, 2008; Rowe, 2007). Studies which included assessment of Motor Skills, found them to be a relative weakness compared to all other skills (Mervis et al., 2001; Mervis & John, 2010; Rowe, 2007). For adults with WS, a different pattern of relative emerged, whereby Socialisation skills typically remained the highest ability area, followed by Daily Living Skills, and Communication skills were the lowest areas of functioning. (Cherniske et al., 2004; Davies et al., 1997; Howlin et al., 1998; Howlin et al., 2010). Comparison of domain scores was not statistically evaluated in some studies (Cherniske et al., 2004; Davies et al., 1997).

Mervis and Klein-Tasman (2000) note that the patterns of relative strengths and weaknesses in adaptive skills fit well with the known cognitive and personality profile of individuals with WS. Relative strengths in Socialisation skills for both children and adults is consistent with personality characteristics of individuals with WS as being extremely friendly and willing to engage in social interactions (Bellugi et al., 2000; Doyle et al., 2004; Meyer-Lindenberg et al., 2006). Their visual-spatial difficulties and problems with motor coordination, balance and muscle tone are likely to contribute to problems independently performing daily living tasks such as dressing, eating, and preparing meals. In addition, living or community skills can include employment skills and understanding and using concepts such as time and money. These tasks demand a certain level of academic and cognitive abilities which are also areas of weakness for individuals with WS.

Variability in Adaptive Functioning

As noted by Howlin et al. (2010), most studies have concentrated on group means, which can obscure information about individual variability. Given the wide range of functioning evidenced by all age groups, it is necessary to consider heterogeneity. It is therefore encouraging that some studies have considered whether individuals deviate from the group mean (Howlin et al., 2010) or whether individuals show different patterns of domain strengths and weaknesses (Mervis et al., 2001). Howlin reported that for the majority of adults in their study, adaptive skills were lower than expected based on IQ level and individual variability was relative small. Using a longitudinal approach, the authors found that at the first assessment adaptive abilities were lower than FSIQ for all participants, whereas at the follow up, nine individual had higher adaptive skills than IQ. Mervis et al. (2001) examined the order of domain scores for each child in their study and reported that 66% of children demonstrated the same order as found at the group level. A limitation is that these studies did not indicate whether these results are based on statistically significant differences either between adaptive and IQ scores or between domain scores.

Is Adaptive Functioning related to Intellectual Ability?

Eleven studies have investigated the relationship between adaptive functioning and intellectual ability (IQ), but there is no clear outcome regarding the relationship. Two studies found no significant difference between mean IQ and overall VABS mean score (Greer et al., 1997; Mervis et al., 2001), while other studies have reported IQ to be significantly lower than adaptive functioning (Fisch et al., 2007; Mervis & John, 2010). Significant positive correlations between IQ and overall adaptive functioning were reported in some studies (Dimitropoulos et al., 2009; Fu, 2012; Howlin et al. 2010; Mervis et al., 2001; Rowe, 2007) with the strength of the relationship varying from a large correlation (e.g., 0.63 reported by Howlin et al., 2010) through to relatively small (0.297 reported by Fu, 2012). Significant positive correlations have also been reported between IQ and the domain of Communication and Daily Living Skills and Community/Personal Living Skills (Davies et al., 1997; Dimitropoulos et al., 2009; Fu, 2012; Howlin et al., 2010; Mervis et al., 2001). Socialisation skills were related to IQ in studies with adults (Davies et al., 1997; Howlin et al., 2010). Motor skills were associated with IQ in a study with children by Mervis et al. (2001), but not with in Fu’s (2012) study with children and adults.

Different intelligence tests have been used across studies (see Table 2 for details), making comparisons difficult. The Wechsler Intelligence Scales were the most commonly employed measure of IQ and a limitation of the Wechsler scales is that they may not be normed low enough to allow sensitive measurement for individuals with very low ID resulting in loss of information about variability in performance for very low functioning individuals and increases the chance of floor effects (Hessl et al., 2009). In comparison, the VABS has a standard score floor of 20, which minimises the chance of floor effects, but makes comparisons between the VABS and Wechsler IQ tests (and other such measures) difficult (Hessl et al., 2009). Comparing results for different adaptive scales can also be problematic, as different scales can vary in how they group together items to create domains. For example, the SIB-R combines Communication and Socialisation abilities into the one domain (Social Interaction and Communication Skills), whereas Vineland scales treat Communication and Socialisation as two separate areas of functioning. Other psychometric issues include the fact that studies have combined IQ results from different IQ batteries, depending upon the age of the participants. There are several psychometric reasons why different tests that purport to measure the same skill may result in significant differences, including floor and ceiling effects, reliability differences and differences in publication dates as tests normed some time ago are found to produce scores that are higher than more recently published tests (Bracken,1988). As noted for adaptive scales, differences can also be found in the way subtests are grouped together to comprise a domain on intelligence tests. For example, the Wechsler Intelligence Scales combine tasks of nonverbal reasoning and visual-spatial reasoning into the one domain of Perceptual Reasoning, while the Differential Ability Scales separates these abilities into a Nonverbal Reasoning domain and a Spatial domain (see Mervis & John, 2010). All of these factors make comparison of studies difficult. Future studies into adaptive functioning would be strengthened by using updated measures with suitable normative data appropriate for all participants.

Adaptive Functioning changes with Chronological Age (CA)

Across a narrow age range (3 to 5 and 4 to 8 years) two studies reported no relationship between chronological age (CA) and overall adaptive functioning abilities (Mervis et al., 2001; Rowe, 2007). These results suggest that children maintain a consistent rate of acquisition of adaptive skills relative to their peers (Mervis et al., 2001). Other studies have reported that for ages ranging from childhood to adolescence, there is evidence that overall adaptive abilities ‘decline’ with increasing CA (Fisch et al., 2007; Mervis & John, 2010). Also, increased chronological age has been found to be significantly associated with lower Communication skills on the VABS (Dimitropoulos et al., 2009) and lower Community Living (Mervis & John, 2010) and Social Interaction/Communication Skills on the SIB-R (Fu, 2012). The two studies which included both children and adults reported different findings regarding the relationship with CA. Di Nuovo and Buono (2011) reporting no relationship between CA and VABS domain scores, however they noted that their findings may have been due to their small sample size (n = 12), whereas Fu’s (2012) study with100 participants reported CA to be negative correlated with Social Interaction and Communication Skills.

This apparent reduction in adaptive functioning abilities with increasing age does not necessarily indicate a loss or decline in functioning. Rather, as noted by Fisch et al. (2007) this is likely to reflect a plateauing of ability, as individuals with WS are not able to continue to gain adaptive abilities at the same rate as their same aged peers (Glaser et al., 2003).

Two studies have conducted longitudinal follow up of participants, with Fisch et al. (2010) re-assessing their participants two years later and found a negligible mean decrease in adaptive functioning indicating relative stability in functioning, at least over a two year period. Howlin et al., 2010 (see also Elison et al., 2010) conducted a 12 year longitudinal follow up of adults and reported significant improvements in their Daily Living and Socialisation skills, suggesting that adaptive skills had improved over that time. It is difficult to compare the results of these studies due to differences in age groups involved and the different follow-up periods used. Further limitations of Howlin et al.'s (2010) study are that while 47 participants took part, only 18 of these had received a positive FISH test since the availability of genetic testing, and the remaining participants had declined to undergo testing. Also, while initial IQ had been assessed on the WAIS-R, follow-up assessment was conducted using the WAIS-III. These limitations can unfortunately reduce the validity of these otherwise very informative studies.

Maladaptive Behaviour

Only three studies included results of Maladaptive behaviours using the VABS Maladaptive Behavior Domain, but not all investigated the potential impact on adaptive functioning. Fisch et al. (2007) reported unusually high levels of Maladaptive behaviours in one third of the children in their study; however they did not report whether these behaviours were related to their adaptive functioning abilities. Howlin et al.'s (2010) longitudinal study found that Maladaptive behaviours in adults were found to decrease significantly over time, with only eight participants showing an increase in problem behaviours from Time 1 to Time 2, but again the relationship with adaptive behaviours was not reported. However, Di Nuovo and Buono (2011) found significant negative correlations between Maladaptive behaviours and each domain on the VABS for 12 participants between 7 and 30 years of age, indicating that higher levels of problem behaviours have the potential to limit adaptive abilities.

Further, using different measures of problem behaviours, Dimitropoulos et al. (2009) reported more frequent problem behaviours, as measured by the Developmental Behavior Checklist (DBC-P), was associated with poorer overall adaptive skills and

Communication, Daily Living, and Socialisation domains, as measured by the VABS. In contrast, Fu (2012) did not find any relationship between adaptive behaviour (SIB-R) and CBCL scores (Total, Internalised, Externalised Problems and Anxious/Depressed scores).

Gender Differences

Four studies (Fisch et al., 2007; Fisch et al., 2010; Fu, 2012; Greer et al., 1997) considered gender differences and none of these studies found evidence of significant differences in adaptive skills between males and females.

Other Factors Related to Adaptive Functioning

Other factors which have been considered for their relationship with adaptive functioning abilities in WS include reduced task persistence and more requests for help when completing tasks for pre-schoolers with WS (Rowe, 2007). Parents rated persistence on the Dimensions of Mastery Questionnaire (DMQ) and the WS children were rated as having lower Social Persistence with Children such as trying hard to make friends (but were not lower on Social Persistence with Adults) and were less persistent with objects or tasks compared to children with Down syndrome, although none of the scores on the DMQ were associated with adaptive functioning scores on the Vineland. On an experimental measure of task mastery, the Individualized Assessment of Mastery Motivation (IAMM), higher task competence was associated with higher adaptive functioning. It should be noted that the tasks of the IAMM involved puzzles, shape sorters and cause-effect toys with all tasks having a number of parts to put together. It is possible that the children with WS would have more difficulty on these sorts of tasks due to their visual-spatial and fine motor difficulties, as compared to children with Down syndrome. Rowe hypothesized that low persistence may account for reduced independence in adulthood.

Higher emotion regulation difficulties were associated with lower adaptive functioning, particularly in the areas of Motor Skills and Personal Living Skills. For this study, Phillips (2008) created a composite measure of emotional regulation by averaging z-scores from the Lability/Negativity subscale of the Emotion Regulation Checklist and the Emotional Control scale from the Behavior Rating Inventory of Executive Functions (BRIEF; Gioia, Isquith, Guy, & Kenworthy, 2000). Studies have also reported that more difficulties with sensory modulation (ability to organize information detected by the senses) measured by parent report (Short Sensory Profile; McIntosh, Miller, Shyu, & Dunn, 1999) was associated with poorer adaptive functioning in children and adolescents (John & Mervis, 2010), while for children and adults, poor visual-motor skills (assessed by the Berry-Buktenica Developmental Test of Visual-Motor Integration, 4th Edition; Beery, 1997) were found to contribute significantly to non-social aspects of adaptive functioning such as motor skills and personal living skills (Fu, 2012).

Discussion

Strengths of the studies which have been completed to-date regarding adaptive functioning ability in WS include the use of well validated, reliable, standardised measures to assess adaptive functioning in both children and adults (e.g., Dimitropoulos et al., 2009; Fisch et al., 2007; Greer et al., 1997; Howlin et al., 1998; Howlin et al., 2010; Mervis et al., 2001). Some studies have included large sample sizes (Davies et al., 1997; Fu, 2012; Howlin et al., 2010). A few studies have looked at individual differences rather than just focusing on group averages (Howlin et al., 2010 and Mervis et al., 2001). Most studies indicated that they used an interview method of administration of adaptive functioning, however, some studies employed a checklist procedure (Fu, 2012; Gosch & Pankau, 1994; Plissart et al., 1994). Conducting an assessment of adaptive functioning by interview, as opposed to having a respondent independently complete the scale, can provide additional clinical information to assist with determining the reliability of responses (Tasse et al., 2012).

A number of limitations have been identified in the research methodology of reviewed studies. Due to the relatively recent advances in genetics, earlier studies relied upon clinical diagnosis of WS. It is encouraging that more recent papers have employed genetic confirmation of WS diagnosis in their research and full genetic analysis would be informative. The use of different tests, either between studies or, in some cases, within the same study and normative data not available for all participant age ranges in the study, are weaknesses of some studies. Use of the most up-to-date valid and reliable measure of adaptive functioning (preferably a scale which provides standardized scores as opposed to age equivalent scores), and IQ should be used where ever possible and appropriate statistical methodology applied.

Directions for future research

As noted by Elison et al. (2010) an understanding of the adaptive behaviour outcomes for individuals with WS is important to ensure appropriate medical, educational, social and employment services can be provided. The presentation of individuals with WS, such as their friendliness and language abilities can mislead professionals and result in an underestimation of the amount of support required to enable participation within the community and increased independence. Dykens and Hodapp (2007) proposed three themes to consider in relation to individual differences within genetic syndromes: “(1) development across the lifespan, (2) gender differences, and (3) other subject and environmental factors” (Dykens & Hodapp, 2007, p. 617). In-line with these themes, further research is needed to increase knowledge about adaptive functioning in the adult population, as four previous studies of adults have reported on groups containing many of the same participants (Davies et al., 1997; Elison et al., 2010; Howlin et al., 1998; Howlin et al., 2010). Studies across children, adolescents and adults would enable further investigation into adaptive functioning across the lifespan in WS. As adolescents complete formal schooling there may be less structure and routine in their lives to continue to develop skills and reduced opportunities for socializing. If this is the case, then, there are important implications regarding ongoing support needs for individuals past the school years. Only four studies in this review considered the potential role of gender differences in adaptive outcomes. While none of these studied reported any evidence of gender difference, this is an area which should be further examined as Porter, Dodd, and Cairns (2009) found a gender difference in behavioural problems in WS with females at a higher risk of developing externalising problems than males. The above points were raised in Martens et al.’s 2008 review, with the recommendation of the use of a wide age range of participants in future WS research and relatively equal numbers of males and females to enable studies to include analyses related to age and gender.

Other within-subject factors, such as intellectual functioning, have been included in many studies and due to the inconclusive results, require ongoing investigation. Lastly, Fu (2012) and Dykens and Hodapp (2007) suggested that environmental and biological factors should be considered in regards to adaptive functioning outcomes. Phillips (2008) noted that measures have concentrated on characteristics of the individual, but have not examined environmental factors such as parenting style. Therefore, future studies should aim to investigate environmental contributions towards adaptive functioning in the WS population.

Conclusion

Adaptive functioning in WS is an area which has received less attention than their intellectual and cognitive functioning. Nevertheless, adaptive functioning is an important area of research due to its relevance to everyday functioning and levels of independence along with the role that is will play in determining severity levels in the diagnosis of intellectual disability. The 18 studies reviewed have provided considerable knowledge to our understanding of adaptive functioning in WS, indicating that these abilities are typically low and that individuals with WS will be more dependent than same-age peers and possibly suggesting lower adaptive functioning for adults. Future research can further increase our knowledge by providing additional information about the predictors and correlates of adaptive functioning in individuals with WS and investigating individual differences.

References

References marked with an asterisk indicate studies included in the systematic review.

American Association on Intellectual and Developmental Disabilities. (2013). *Definition of Intellectual Disabilities.* Retrieved from http://aaidd.org/intellectual-disability/definition#.U5hCGaaKCpo.

American Psychiatric Association. (2000). *Diagnostic and statistical manual of mental disorders (*4th ed., Text Rev.). Washington, DC: Author.

American Psychiatric Association. (2013). *Diagnostic and statistical manual of mental disorders* (5th ed.). Washington, DC: Author.

Antonell, A., Del Campo, M., Magano, L.F., Kaufmann, L., Martinez de la Iglesia, J., Gallastegui, F., Flores, R., Schweigmann, U., Fauth, C., Kotzot, D., Perez-Jurado, L.A. (2010). Partial 7q11.23 deletions further implicate GTFI and GTF2IRD1 as the main genes responsible for the Williams-Beuren syndrome neurocognitive profile. *Journal of Medical Genetics, 47,* 312-320.

Beery, K.E. (1997). The Beery-Buktenica Developmental Test of Visual-Motor Integration: Administration, Scoring, and Teaching Manual (4th Revised Edition). Parsippany, NJ: Modern Curriculum Press.

Bellugi, U., Lichtenberger, L., Jones, W., Lai, Z., & St. George, M. (2000). The neurocognitive profile of Williams syndrome: A complex pattern of strengths and weaknesses. *Journal of Cognitive Neuroscience, 12,* 7-29.

Bracken, B.A. (1988). Ten psychometric reasons why similar tests produce dissimilar results. *Journal of School Psychology, 26*, 155-166.

Bruininks, R.H., Woodcock. R.W., Weatherman, R.F., & Hill, B.K. (1996). *Scales of Independent Behavior – Revised.* Itasca, IL: Riverside Publishing Company.

*Cherniske, E.M., Carpenter, T.O., Klaiman, C., Young, E., Bregman, J., Insogna, K., Schultz, R.T., & Pober, B.R. (2004). Multisystem study of 20 older adults with Williams syndrome. *American Journal of Medical Genetics, 131A*, 255-264.

Crawford, N.A., Edelson, L.R., Skwerer, D.P., & Tager-Flusberg, H. (2008). Expressive language style among adolescents and adults with Williams syndrome. *Applied Psycholinguistics, 29,* 585-602.

*Davies, M., Howlin, P., & Udwin, O. (1997). Independence and adaptive behavior in adults with Williams syndrome. *American Journal of Medical Genetics, 70*, 188-195.

*Dimitropoulos, A., Ho, A.Y., Klaiman, C., Koenig, K., Schultz, R.T. (2009). A comparison of behavioral and emotional characteristics in children with autism, Prader-Willi syndrome, and Williams syndrome. *Journal of Mental Health Research in Intellectual Disabilities, 2,* 220-243.

*Di Nuovo, S., & Buono, S. (2011). Behavioral phenotypes of genetic syndromes with intellectual disability: Comparison of adaptive profiles. *Psychiatry Research, 189,* 440-445.

Dixon, D.R. (2007). Adaptive behavior scales. In J.L. Matson (Ed.), *International Review of Research in Mental Retardation*: Vol 34. Handbook of Assessment in Persons with Intellectual Disability (pp. 99-140). Oxford, England: Elsevier.

Doyle, T.F., Bellugi, U., Korenberg, J.R., & Graham, J. (2004). "Everybody in the world is my friend": Hypersociability in young children with Williams syndrome. *American Journal of Medical Genetics, 124A,* 263-273.

Dykens, E.M., & Hodapp, R.M. (2007). Three steps toward improving the measurement of behavior in behavioral phenotype research. *Neuropsychiatric Genetic Syndromes, 16,* 617-630.

Edgin, J.O., Pennington, B.F., & Mervis, C.B. (2010). Neuropsychological components of intellectual disability: the contributions of immediate, working and associative memory. *Journal of Intellectual Disability Research, 54*, 406-417.

*Elison, S., Stinton, C., & Howlin, P. (2010). Health and social outcomes in adults with Williams syndrome: Findings from cross-sectional and longitudinal cohorts. *Research in Developmental Disabilities, 31*, 587-599.

*Fisch, G.S., Carpenter, N., Howard-Peebles, P.N., Holden, J.J.A., Tarleton, J., Simensen, R., Nance, W. (2007). Research review: Studies of age-correlated features of cognitive-behavioral development in children and adolescents with genetic disorders. *American Journal of Medical Genetics Part A, 143A,* 2478-2489.

*Fisch, G.S., Carpenter, N., Howard-Peebles, P.N., Holden, J.J.A., Tarleton, J., & Simensen, R. (2010) The course of cognitive-behavioral development in children with FMR1 Mutation, Williams-Beuren syndrome, and Neurofibromatosis Type 1: The effect of gender. *American Journal of Medical Genetics Part A, 152A*, 1498-1509.

*Fu, J.T. (2012). The association of intelligence, visual-motor functioning, and personality characteristics with adaptive behavior in individuals with Williams syndrome. (Order No. 3524353, Alliant International University). *ProQuest Dissertations and Theses,* 117. Retrieved from http://search.proquest.com/docview/1080530412?accountid=12219. (1080530412).

Gioia, G.A., Isquith, P.K., Guy, S.C., Kenworthy, L. (2000). *Behavior Rating Inventory of Executive Function*. Lutz, FL: Psychological Assessment Resources.

Glaser, B., Hessl, D., Dyer-Friedman, J., Johnston, C., Wisbeck, J., Taylor, A., & Reiss, A. (2003). Biological and environmental contributions to adaptive behaviour in Fragile X syndrome. *American Journal of Medical Genetics, 117A,* 21-29.

*Gosch, A., & Pankau, R. (1994). Social-emotional and behavioral adjustment in children with Williams-Beuren syndrome. *American Journal of Medical Genetics, 53,* 335-339.

*Greer, M.K., Brown III, F.R., Shashidhar Pai, G., Choudry, S.H., & Klein, A.J. (1997). Cognitive, adaptive, and behavioral characteristics of Williams syndrome. *American Journal of Medical Genetics (Neuropsychiatric Genetics), 74*, 521-525.

Harries, J., Guscia, R., Kirby, Nettelbeck, T., Taplin, J., & MacLean, W.E. (2005). Support needs and adaptive behaviors. *American Journal on Mental Retardation, 110,* 393-404.

Harrison, P.L. (1987). Research with adaptive behavior scales. *The Journal of Special Education, 21,* 37-68.

Harrison, P.L., & Boney, T.L. (2002). Best practices in the assessment of adaptive behavior. In A. Thomas & J. Grimes (Eds), *Best Practices in School Psychology-IV* (pp. 1167-1179). Washington, DC: National Association of School Psychologists.

Hessl, D., Nguyen, D.V., Green, C., Chavez, A., Tassone, F., Hagerman, R.J., Senturk, D., Schneider, A., Lightbody, A., Reiss, A.L., & Hall, S. (2009). A solution to limitations of cognitive testing in children with intellectual disabilities: The case of Fragile X syndrome. *Journal of Neurodevelopmental Disorders, 1,* 33-45.

Hogan, T.P. (2003). *Psychological Testing: A Practical Introduction.* New York: John Wiley & Sons.

*Howlin, P., Davies, M., Udwin, O. (1998). Cognitive functioning in adults with Williams syndrome. *Journal of Child Psychology and Psychiatry, 39,* 183-189.

*Howlin, P., Elison, S., Udwin, O., & Stinton, C. (2010). Cognitive, linguistic and adaptive functioning in Williams syndrome: Trajectories from early to middle adulthood. *Journal of Applied Research in Intellectual Disabilities, 23,* 322-336.

Jarrold, C., Baddeley, A.D., & Hewes, A.K. (1998). Verbal and non-verbal abilities in the Williams syndrome phenotype: Evidence for diverging developmental trajectories. *Journal of Child Psychology and Psychiatry, 39*, 511-523.

*John, A.E., & Mervis, C.B. (2010). Sensory modulation impairments in children with Williams syndrome. *American Journal of Medical Genetics Part C (Seminars in Medical Genetics), 154C*, 266-276.

Lowery, M.C., Morris, C.A., Ewart, A., Brothman, L.J., Zhu, X.L., Leonard, C.O., Carey, J.C., Keating, M., Brothman, A.R. (1995). Strong Correlation of Elastin

Deletions, Detected by FISH, with Williams Syndrome: Evaluation of 235 Patients. *American Journal of Human Genetics, 57*, 49-53.

Martens, M.A., Wilson, S.J., & Reutens, D.C. (2008). Research review: Williams syndrome: A critical review of the cognitive, behavioral, and neuroanatomical phenotype. *Journal of Child Psychology and Psychiatry, 49*, 576-608.

McGrew, K. (2012).What is the typical IQ and adaptive behavior correlation? (Brief Report No. 11): Retrieved from IAP Applied Psychometrics website: http://www.ipscorner.com/2012/02/iap-applied-psychometrics-101-brief.html

McIntosch, D.N., Miller, L.J., Shyu, V., & Dunn, W. (1999). Short sensory profile. NY: The Psychological Corporation.

*Mervis, C.B., & John, A.E. (2010). Cognitive and behavioral characteristics of children with Williams syndrome. *American Journal of Medical Genetics Part C (Seminars in Medical Genetics), 154C*, 229-248.

Mervis, C.B., & Klein-Tasman, B.P. (2000). Williams syndrome: Cognition, personality, and adaptive behavior. *Mental Retardation and Developmental Disabilities Research Reviews, 6,* 148-158.

*Mervis, C.B., Klein-Tasman, B.P., & Mastin, M.E. (2001). Adaptive behavior of 4- through 8-year old children with Williams syndrome. *American Journal on Mental Retardation, 106,* 82-93.

Mervis, C.B., & Morris, C.A. (2007). Williams syndrome. In M.M. Mazzocco, & J.L. Ross (Eds.), *Neurogenetic developmental disorders: Variation of manifestation in childhood* (pp.199-262). Cambridge, MA: MIT Press.

Meyer-Lindenberg, A., Mervis, C.B., & Berman, K.F. (2006). Neural mechanisms in Williams syndrome: A unique window to genetic influences on cognition and behaviour. *Nature Reviews Neuroscience, 7,* 380-393.

Pezzini, G., Vicari, S., Voltera, V., Milani, L., & Ossella, M.T. (1999). Children with Williams syndrome: Is there a single neuropsychological profile? *Neuropsychologia, 15*, 141-155.

*Phillips, K.D. (2008). Emotion regulation and dysregulation in children and adolescents with Williams syndrome (Doctoral dissertation). Retrieved from ProQuest database. (304453044).

Philofsky, A.D. (2006). Pragmatic language profiles in autism and Williams syndrome (Doctoral dissertation). Retrieved from ProQuest database. (305352531)

*Plissart, L., Borghgraef, M., Volcke P., Van den Berghe, H., & Fryns, J.P. (1994). Adults with Williams-Beuren syndrome: Evaluation of the medical, psychological and behavioral aspects. *Clinical Genetics, 46,* 161-167.

Porter, M.A. & Coltheart, M., (2005). Cognitive heterogeneity in Williams syndrome. *Developmental Neuropsychology, 27,* 275-306.

Porter, M.A., Dobson-Stone, C., Kwok, J.B.J., Schofield, P.R., Beckett, W., & Tassabehji, M. (2012). A role for transcription factor GTF2IRD2 in executive function in Williams-Beuren syndrome. *PLoS ONE, 7,* e47457. DOI: 10.1371/journal.pone.0047457.

Porter, M.A., Dodd, H., & Cairns, D. (2009). Psychopathological and behaviour impairments in Williams-Beuren syndrome: The influence of gender, chronological age, and cognition. *Child Neuropsychology, 15,* 359-374.

Rothman, K.J. (1990). No adjustments are needed for multiple comparisons. *Epidemiology, 1,* 43-46.

*Rowe, M.L. (2007). Mastery motivation in young children with Williams syndrome or Down syndrome (Order No. 3267110, University of Louisville). *ProQuest Dissertations and Theses,* 131. Retrieved from http://search.proquest.com/docview/304837706?accountid=12219. (304837706).

Sattler, J.M. (2008). *Assessment of children: Cognitive foundations* (5th ed.). La Mesa, CA: J.M. Sattler.

Schalock, R.L. (2004). The emerging disability paradigm and its implications for policy and practice. *Journal of Disability Policy Studies, 14,* 204-215.

Sparrow, S., Balla, D., & Cicchetti, D. (1984). *Vineland Adaptive Behavior Scales. Survey Forms Manual.* Circle Pines, MN: American Guidance Services.

Sparrow, S.S., Cicchetti, D.V., & Balla, D.A. (2005). *Vineland Adaptive Behavior Scales – Second Edition.* Circle Pines, MN: AGS Publishing.

Strømme, P., Bjørnstad, P.G., Ramstad, K. (2002). Prevalence estimation of Williams syndrome. *Journal of Child Neurology, 17,* 269-271.

Tassabehji, M., Metcalfe, K., Karmiloff-Smith, A., Carette, M.J., Grant, J., Dennis, N., Reardon, W., Splitt, M., Read, A.P., & Donnai, D. (1999). Williams syndrome: Use of chromosomal microdeletions as a tool to dissect cognitive and physical phenotypes. *American Journal of Human Genetics, 64*, 118-125.

Tasse, M.J., Schalock, R.L., Balboni, G., Bersani, H., Borthwick-Duffy, S.A., Spreat, S., Thissen, D., Widaman, K.F., & Zhang, D. (2012). The construct of adaptive behavior: Its conceptualization, measurement, and use in the field of intellectual disability. *American Journal on Intellectual and Developmental Disabilities, 117*, 291-303.

Thompson, J.R., McGrew, K.S., & Bruininks, R.H. (2002). Pieces of the Puzzle: Measuring the Personal Competence and Support Needs of Persons with Intellectual Disabilities, *Peabody Journal of Education, 77,* 23-39.

Venn, J.J. (2007). *Assessing students with special needs* (4th ed.). Upper Saddle River, NJ: Pearson Education.

Williams, J., Barrett-Boyes, B., & Lowe, J. (1961). Supravalvular aortic stenosis. *Circulation, 24,* 1311-1318.

Adaptive functioning in Williams syndrome and its relation to demographic variables and family environment

Gabrielle M. Brawn[1] and Melanie A. Porter[1]

Psychology Department, Macquarie University, Sydney, NSW, Australia

Key words: adaptive functioning, family environment, Williams syndrome

Correspondence should be sent to Melanie A. Porter, Psychology Department, Macquarie University, Sydney, Australia 2109. Phone: +61 2 9850 6768, Fax: +61 2 9850 8062, E-mail: melanie.porter@mq.edu.au

Abstract

This study used the Vineland Adaptive Behavior Scale - 2nd Edition (Vineland II) to investigate adaptive functioning in a group of 30 children and adults with Williams syndrome (WS). The study had three aims: 1) to profile adaptive functioning in WS; 2) to investigate the relationship between adaptive functions and the demographic variables of gender, chronological age, and IQ (with IQ assessed using the Woodcock-Johnson – 3rd Edition, Tests of Cognitive Abilities or WJ III COG); and 3) to investigate the relationship between levels of adaptive functioning and family environment characteristics (the latter measured using the Family Environment Scale or FES). In line with predictions: 1) there was extensive variability in overall levels of adaptive functions and in relative strengths and weaknesses at the domain and subdomain levels; 2) while neither gender nor IQ were significantly related to adaptive skills, Communication skills at the Domain level and Interpersonal Relationship skills at the subdomain level failed to make appropriate gains relative to same aged peers, 3) adaptive functioning was significantly related to family environment, in particular, participation and interest in Intellectual-Cultural and Active-Recreational activities, and the use of set rules and routines within the family (Control). Implications for the diagnosis of intellectual disability, access to funding, referral to appropriate support services and educational and vocational plans are discussed.

Introduction

While research into the cognitive and intellectual functioning of individuals with Williams syndrome (WS) has received a lot of attention to date, there has been less research into adaptive functioning and any factors which may impact on the development of these skills.

Adaptive functioning (or adaptive behaviour) refers to the learned conceptual, social and practical skills performed by an individual in their day-to-day lives (Tasse et al., 2012). Information about adaptive functioning is important to guide early intervention, educational and employment experiences and would benefit families and carers as they endeavour to support individuals with WS to live as independently as possible. For example, as adaptive functioning measures 'real-life' skills, it can be a good indicator of the level of support an individual will require in learning situations, suitable employment and recreational options, and the assistance they may need in managing legal decisions, health care, transportation and finances (American Psychological Association [APA], 2013).

Understanding adaptive functions also has implications for the diagnosis and classification of intellectual disability (ID), eligibility for funding support and services and in determining the least restrictive environment and level of supervision required for individuals with WS (Dixon, 2007). This is particularly pertinent with the recent revision of the Diagnostic and Statistical Manual of Mental Disorders [Fifth Edition (DSM-5), published by the American Psychiatric Association (APA, 2013)], where adaptive functioning has become a more prominent component of the diagnosis and classification of ID. While deficits in both intellectual and adaptive functioning continue to be the essential features of ID, the level of severity of ID (mild, moderate, severe, or profound) is no longer based on IQ scores, but will now be defined by adaptive functioning abilities across the three domains of conceptual (academic), social and practical abilities (APA, 2013).

The scant literature available regarding adaptive functioning in WS remains inconclusive in terms of relative strengths and weaknesses of adaptive skills and in terms of the relationship between adaptive functioning and gender, chronological age or level of intellectual functioning. Also, no research to date has explored how family characteristics, such as family attitudes and family culture might influence daily independence. This study aimed to investigate adaptive functioning in WS, exploring the profile of adaptive functioning skills and how this relates to demographic variables and family characteristics. As WS is commonly associated with extensive variability in cognitive and behavioural skills (Davies, Howlin, & Udwin, 1997; Jarrold, Baddeley, & Hewes, 1998; John & Mervis, 2010; Porter & Coltheart, 2005), group patterns were explored, as well as individual profiles of strength and weakness.

Williams syndrome

Williams syndrome, also referred to as Williams-Beuren syndrome, is a genetic disorder resulting from a hemizygous microdeletion on chromosome 7 at the location 7q11.23. The typical deleted region involves around 26 genes, including the elastin gene (ELN), and results in cardiovascular, connective tissue and neurodevelopmental deficits. The prevalence of WS is estimated to be approximately 1 in 7,500 births (Strømme, Bjørnstad, & Ramstad, 2002). Characteristic features of WS include dysmorphic facial features, small stature, hyperacusis, transient-neonatal hypercalcemia and a specific cognitive and behavioural phenotype. Cognitively, the WS profile generally includes relative strengths in verbal short-term memory, some aspects of language (receptive vocabulary) and face recognition and relative weaknesses in visuo-spatial skills (see reviews by Martens, Wilson, & Reutens, 2008; Meyer-Lindenberg, Mervis, & Berman, 2006). However, there is some variability in the cognitive strengths and weaknesses of people with WS (Jarrold et al., 1998; Pezzini, Vicari, Voltera, Milani, & Ossella, 1999; Porter & Coltheart, 2005; Tassabehji et al., 1999). Behaviourally, individuals with WS are noted for their outgoing and extremely friendly personalities and have been described as hypersociable (Jones et al., 2000). At the same time, they experience higher levels of generalised anxiety and specific phobias (Dodd & Porter, 2009; Einfeld, Tonge, & Florio, 1997; Kennedy, Kaye, & Sadler, 2006) and have difficulty forming and especially maintaining friendships with same age peers (Dimitropoulos, Ho, Klaiman, Koenig, & Schultz, 2009; Gosch & Pankau, 1994).

The recognised cognitive and behavioural phenotype of WS has connotations for adaptive functioning outcomes. For example, their friendly, outgoing personalities may be expected to result in higher scores on scales measuring social skills, while difficulties in the area of maintaining friendships may perhaps be expected to negatively influence these scores. Likewise, reported strengths in verbal abilities may mean higher scores on the communication scales of adaptive functioning. Cognitive variability may also mean that not all individuals with WS will display the same profile of strengths and weaknesses in adaptive skills or the same overall level of adaptive abilities.

Intellectual Disability in WS: IQ and Adaptive Functions

Intellectual functions typically lie within the range of a mild to moderate disability (with IQ scores typically within the range of 50 to 70)[2] (Bellugi, Lichtenberger, Jones, Lai, & St. George, 2000; Howlin, Davies, & Udwin, 1998; Martens et al., 2008). WS individuals display variable performances on IQ tests, however, with some individuals reported to be functioning at the level of a severe intellectual impairment (up to four standard deviations below the population mean) or, conversely, within one standard deviation of the population mean and in the average range (see Mervis & John, 2010 for a review). This indicates that while the majority of individuals with WS do have an intellectual impairment, the level of severity varies widely.

IQ profile scores also seem to vary in WS. There have been reports of significant differences between verbal and nonverbal (spatial) intellect in WS (see Martens et al., 2008 for a review), yet other studies have failed to find a significant difference between verbal and nonverbal domains (e.g., Bellugi et al., 2000). With regard to the former, in some studies verbal intellect is reported as being significantly higher than nonverbal intellect (e.g., Searcy et al., 2004). There are also a few reports of significantly higher nonverbal than verbal intellect (Howlin, Elison, Udwin, & Stinton, 2010; Searcy et al., 2004). While there have been assertions of a distinct WS intellectual profile in WS, with strengths in verbal skills and weaknesses in nonverbal (spatial) skills (Mervis & Klein-Tasman, 2000; Morris, 2010), most of the early studies tended to focus on group profiles or group averages, thus masking individual variability. It has now been recognised that not all individuals with WS demonstrate the same cognitive level or intellectual strengths and weaknesses. This highlights the need to go beyond group averages and to explore individual profiles of ability. It also

[2] According to the Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition, Text Revision (DSM-IV), American Psychiatric Association (APA, 2000).

highlights the need to explore why this variability occurs. Variability is likely to reflect inherent and environmental factors.

Adaptive functioning in WS

The most commonly used measure to assess adaptive functioning in WS has been the Vineland Adaptive Behavior Scales (VABS; Sparrow, Balla, & Cicchetti, 1984). The VABS is a well-known and widely used instrument to measure adaptive functioning across a wide range of developmental disorders such as autism, Down syndrome or Fragile X syndrome (see Dixon, 2007 for a review of published studies). However, the normative data available for the VABS is limited to the age of 18 years 11 months (Sparrow et al., 1984). In addition to the VABS, the Scales of Independent Behavior – Revised (SIB-R; Bruininks, Woodcock, Weatherman, & Hill, 1996) with norms which cover the age range from infants to 80 years has been used to measure adaptive functioning in three studies on WS.

Adaptive scales, like IQ tests, render a standard score with a mean of 100 and standard deviation of 15. Using this metric, studies have reported that the majority (typically around 75%) of individuals with WS have adaptive functioning levels at an impaired or low level. That is, more than two standard deviations below the population mean (and a standard score of less than 70) (Mervis & Klein-Tasman, 2000). Typically, the mean level of adaptive ability falls within the mild to moderate range, which is consistent with the majority of studies on IQ levels (Greer, Brown, Pai, Choudry, & Klein, 1997). However, as with IQ, there is evidence of heterogeneity, with some individuals functioning at an extremely low level, while others are functioning at a chronologically age appropriate (average) level. This again highlights the need to consider individuals case-by-case. Extensive intra-individual variability is also commonly reported in WS, which indicates the need to go beyond global measures of adaptive functioning and to explore strengths and weaknesses at the domain and subdomain level.

Strengths and weaknesses in adaptive functioning

Adaptive functioning is considered a multidimensional construct, and while researchers continue to report a unified composite score, the trend has become to evaluate differences on the specific domain scores (Dixon, 2007). This is particularly relevant to consider for WS with its peaks and troughs in ability. Some patterns reported in specific domains of adaptive functioning, such as Social, Communication or Daily Living Skills generally fit with the behavioural, physical and cognitive phenotype of WS, at least at the group level. From the available studies of adaptive functioning in WS, children and adolescents were rated highest on their Socialisation skills compared to the domains of Communication and Daily Living skills (Dimitropoulos et al., 2009, Fisch et al., 2007, Greer et al., 1997; Mervis, Klein-Tasman, & Mastin, 2001). Of note, studies involving adults with WS have found slightly different profiles of adaptive functioning to studies of children with WS. While Socialisation skills remain a relative strength in adults, Daily Living skills become the next highest skill, and Communication skills become the weakest area of functioning (Cherniske et al., 2004; Davies et al., 1997; Howlin et al., 1998, Howlin et al., 2010). These differences were confirmed as statistically significant in two of these studies (Howlin et al., 1998; Howlin et al., 2010).

A relative strength in Socialisation skills is consistent with the personality profile of individuals with WS being extremely friendly and willing to engage in social interactions (Bellugi et al., 2000; Meyer-Lindenberg et al., 2006). Similarly, a relative strength in Communication skills has been observed in children and adolescents with WS and is consistent with reports of strengths in verbal abilities, including verbal intellect in WS (Mervis et al., 2001). Also, difficulties with spatial and motor functioning identified in WS are thought to result in lower self-care and independence skills reflected by the Daily Living Skills domain on the VABS (Fu, 2012; Gosch & Pankau, 1994; Mervis & Klein-Tasman, 2000).

Only two studies (Howlin et al., 2010; Mervis et al., 2001) have explored individual adaptive functioning profiles in WS. One study (Mervis et al., 2001) examined the pattern of domain scores for each individual child and reported that the same pattern (Socialisation higher than Communication and Communication higher than Daily Living Skills) was found for 66% of the children in their study, however, it is not known whether this pattern of differences was statistically significant for the majority of individuals. One other study on adults with WS reported that individual variation was relatively small (Howlin et al., 2010).

In summary, the overall level of adaptive functioning in WS is typically found to be below the level expected for an individual's chronological age, with general mean level of functioning ranging from a mild to moderate level of impairment. Socialisation appears to be a relative strength for both children and adults; however there may be some age related differences, as Communication skills are a relative weakness and the lowest area of functioning for adults.

Maladaptive Behaviour in WS

Maladaptive behaviours have the potential to impact on individuals' adaptive abilities by limiting their capacity to participate in daily activities and benefit from education, training and community inclusion (de Bildt, Sytema, Kraijer, Sparrow, & Minderaa, 2005; Di Nuovo & Buono, 2011). While the SIB-R and VABS (the two most commonly used measures of adaptive function in WS) include maladaptive scales, not many studies have utilised them in their research to explore how these problem behaviours may relate to overall levels of adaptive function. Of those that did, Di Nuovo and Buono (2011) reported that high levels of problem behaviours as measured by the VABS were associated with poorer Communication, Daily Living Skills and Socialisation abilities in a group of WS individuals spanning children to adults. Similarly, Phillips (2008) reported that once IQ was accounted for, poor emotional control was associated with lower overall adaptive skills, motor skills and personal living skills on the SIB-R and Dimitropoulos et al. (2009) found that a higher total behaviour problem score (using the Developmental Behavior Checklist; Einfeld & Tonge, 2002) was associated with lower overall adaptive skills and Communication, Daily Living and Socialisation skills on the VABS. One study with a wide age range of children and adults (12-53 years) with WS failed to find a significant relationship between internalizing problems (measured using the Child Behaviour Checklist; Achenbach & Rescorla, 2001) and adaptive functioning (Fu, 2012). These discrepant findings may be due to the use of different measurement tools to assess problem behaviours and adaptive functioning across studies, which makes comparisons of studies difficult.

The relationship between adaptive functions and the variables of gender, chronological age, and IQ

Adaptive functions and gender. Of the four studies to date which have explored whether there are significant differences in adaptive functioning for males and females with WS, no study has found any significant gender differences (Fisch et al., 2007, Fisch et al., 2010; Fu, 2012; Greer et al., 1997). In terms of maladaptive behaviours, of note, Porter, Dodd, and Cairns (2009) reported that females in their study were at a higher risk of developing externalising problems than males. No other study has reported gender differences in maladaptive behaviour in WS.

Changes in adaptive functioning with age. Most previous studies have explored adaptive functioning in WS within either a younger age range (including children or children and adolescents) or with adult populations. No changes in adaptive abilities with age were identified for children across narrow age ranges, such as from 3 to 5 years of age (Rowe, 2007) or from 4 to 8 years of age (Mervis et al., 2001). However, across larger age ranges spanning childhood and adolescence, there has been evidence of a significant negative relationship between overall adaptive abilities and chronological age (Fisch et al., 2007; Mervis & John, 2010). Also, increased chronological age has been found to be associated with lower Community Living (Mervis & John, 2010) and Social Interaction/Communication Skills on the SIB-R (Fu, 2012) and Communication skills on the VABS (Dimitropoulos et al., 2009). Fisch et al. (2007) proposed that the inverse relationship of decreasing adaptive skills with increasing chronological age is not necessarily associated with a decline in functioning, but may rather relate to a plateauing in development. That is, their development is more gradual than typically developing children and, as they get older, the gap between their ability and their chronological age widens compared to typically developing individuals of the same age. In their longitudinal follow up two years later, Fisch et al. (2010) found that retest adaptive functioning scores did not change significantly from their initial values, confirming that there does not appear to be a loss of skill over time. In contrast to Fisch et al., Howlin et al.'s (2010) longitudinal study reported that, while individuals were functioning below the level expected for their chronological age, their overall adaptive abilities and Daily Living and Socialisation skills appeared to improve with age. This discrepancy may be due to the difference in ages of the participants and differences in the follow up times. Fisch et al.'s participants were originally aged 3 to 16 years and followed up two years later. Perhaps this may not have been long enough to detect significant changes. Howlin et al.'s participants were older (25 to 49 years) and followed up after a longer time period of 12 years.

Only two studies to date have included participants across a wide age range spanning from children to adults. Di Nuovo and Buono (2011) studied individuals with WS aged 7 to 30 years, and Fu's (2012) study spanned the ages of 12 to 53 years. Di Nuovo and Buono (2011) found no significant improvements or decreases in adaptive abilities with age on an Italian version of the VABS; they explored overall adaptive abilities and also looked at the domain level. Fu (2012) utilised a much larger sample in a cross-sectional study of 100 WS individuals and found that for older participants, their Social and Communication skills were significantly lower than that of younger participants. Fu concluded that children and adults may have different adaptive profiles, particularly in the area of Communication skills, and consistent with reports by Howlin et al. (1998) and Cherniske et al. (2004) who found significant impairments especially in the Communication domain on the VABS for older individuals with WS. This may represent evidence of a deviant developmental trajectory in WS as proposed by Donnai and Karmiloff-Smith (2000). It is worth noting, however, that others have failed to find any evidence of a decline in language skills with age (e.g. Howlin et al.'s 2010 twelve year longitudinal follow-up study). Furthermore, Howlin et al. (2010) reported no significant difference in FSIQ or VIQ over time, with the majority of individuals having a higher VIQ than PIQ score at both times (see also Searcy et al., 2004).

While performance on tests of receptive vocabulary is a relative strength, other aspects of their language abilities, such as poor pragmatic skills, may explain difficulties with adaptive Communication skills with age (Brock, 2007). In addition, adaptive scale items shift emphasis from receptive and expressive language abilities towards an increasing demand on written communication abilities with age (Mervis et al., 2001). Written language skills rely on cognitive, academic and motor abilities, areas in which individuals with WS have difficulty.

The Relation between adaptive functioning and IQ. Research on typically developing populations to date suggests that adaptive functioning and IQ should be considered as related yet distinct constructs (Harrison & Boney, 2002; Keith, Fehrmann, Harrison, & Pottebaum, 1987), with relationships between adaptive functioning and IQ varying widely from low through to moderate (Harrison & Boney, 2002). Nevertheless, there has been some evidence that the relationship between IQ and adaptive functioning is stronger for individuals with intellectual disability (Liss et al., 2001; Sattler, 2001), but this does not always appear to be the case (Harrison, 1987).

In the WS literature, different methods have been used to investigate the association between intellectual functioning and adaptive functioning and findings remain inconclusive regardless of the method used. The first method has been to look at whether there is a significant difference between overall IQ and adaptive functioning scores. The other method has been to look at correlations between IQ scores and adaptive functioning scores.

In relation to the first method, some studies have reported no significant difference between IQ and adaptive functioning (Greer et al., 1997; Mervis et al., 2001) while other studies have reported higher (Fisch et al., 2007) or lower (Howlin et al., 1998; Mervis & John, 2010) levels of adaptive functioning compared to IQ. In relation to the second method, reports have varied from small to large correlations between adaptive skills and IQ in WS. Some of the variable findings in terms of relationship between IQ and adaptive functioning may be due to the specific measures employed (Keith et al., 1987).

At a case by case level in a cross-sectional and longitudinal study, Howlin et al. (2010) noted FSIQ to be higher than the global adaptive functioning score (ABC) in the majority (90%) of individuals in the cross-sectional study and in all individuals of the longitudinal study at Time 1. The reverse pattern (of ABC score higher than FSIQ) was found in nine individuals (10%) in the cross-sectional study and in nine individuals (20%) in the 12 year follow-up. As the same participants took part in these studies, it is possible that these nine individuals are the same in both studies, but this information was not reported. The mean gap between FSIQ and ABC score was reported to be narrower in those aged over 30 years compared to younger participants (less than 30 years of age), which the authors suggested indicates gains in adaptive functioning with age (Howlin et al., 2010). It is not known how many, if any, of the reported differences are statistically significant and it is unclear whether measurement error was taken into account.

Correlational studies have ranged from reports of a small correlation between IQ and adaptive functioning scores in WS (e.g., .297 reported by Fu, 2012) to large correlations (e.g., .63 reported by Howlin et al., 2010). Correlations have also been reported between FSIQ and domain scores (Davies et al., 1997; Dimitropoulos et al., 2009; Fu, 2012; Howlin et al., 2010; Mervis et al., 2001). Correlations between FSIQ and the Communication domain on the VABS ranged from .42 to .70. Daily Living Skills ranged from .34 to .59, and Socialisation was from .37 to .56. Two studies (Davies et al., 1997 and Howlin et al., 2010) reported significant positive correlations between FSIQ and all three domains of the VABS (both studies involved the same participants). Fu (2012) also found significant positive correlations between FSIQ and three cluster scores of the SIB-R (Social/Communication, Personal Living, and Community Living).

Howlin et al. (2010) was one of the few studies to go beyond FSIQ and to explore the relationship between verbal intellect (VIQ) and nonverbal intellect (PIQ) with adaptive functioning. Significant positive correlations of level of adaptive functioning and verbal (.59) and nonverbal (.57) intellect were reported.

Using regression models, Fu (2012) reported that IQ accounted for significant variance in adaptive skills. A short-form estimate of IQ was used, focusing on verbal ability. Fu reported that verbal IQ accounted for the largest amount of unique variance (15.6%) in the Social Interaction and Communication domain, while in the Personal Living Skills domain visual-motor functioning was the only significant predictor of adaptive functioning. This further supports the view that IQ and adaptive functioning are related yet distinct abilities.

The methodological differences between all of the above studies make direct comparisons and interpretations across studies difficult. Most studies have used either the VABS or the SIB-R to assess adaptive functioning in WS and these two measures differ in their domain/cluster constructs, as the VABS has separate Communication and Socialisation domains, while the SIB-R combines these skill areas into a single Social Interaction and Communication score. A wide variety of measures have been employed to assess IQ, both between studies and within the same study and discrepancies can exist between different tests which are designed to measure the same skill (Bracken, 1988). All of these differences make it difficult to compare results. Nevertheless, the relationship between IQ and adaptive functioning is clinically relevant, as it would be important to know whether intellectual or, indeed, more specific cognitive skills are predictive of a WS individual's ability to attain functional independence and whether it is reasonable to expect higher levels of adaptive abilities in WS despite a low level of intellectual functioning.

Adaptive functions and the family context

No WS study to date has explored environmental contributions of family attitudes and family culture to an individual's adaptive functioning capabilities. In children with intellectual disability more generally, however, more cohesive and harmonious family functioning has been found to relate to more positive socio-emotional functioning (Mink, Nihira, & Meyers, 1983), fewer behaviour problems (Warfield, 1995) and these supportive family relationships during early childhood seem to predict positive social relationships in middle childhood (Howell, Hauser-Cram, & Kersh, 2007). Similarly, Hauser-Cram et al. (1999) suggest that the family can influence a child's development through family characteristics, including patterns of interactions among family members.

For young children with Down syndrome, growth in the adaptive ability areas of Communication, Daily Living Skills and Socialisation as measured by the VABS over a five year period was significantly predicted by family environment factors such as family cohesion (as measured by the Family Adaptability and Cohesion Evaluation Scale) and mother-child interaction (using an observational scale, the Nursing Child Assessment Teaching Scale; Hauser-Cram et al., 1999). The authors concluded that even though Down syndrome is biologically based, understanding the family environment in relation to their adaptive development is important (Hauser-Cram et al., 1999).

One study also considered the relationship between environmental factors and adaptive functioning in Fragile X syndrome using the VABS and the Home Observation for Measurement of the Environment or HOME (Bradley, 1993) interview and observation (Glaser et al., 2003). Glaser et al. found that the home environment (as measured by the HOME scale) was a significant predictor of adaptive functioning in males with Fragile X, but not females (Glaser et al., 2003). Characteristics of the home environment were not considered individually as a mean overall HOME score was utilised, so it is not clear which aspects of the home environment were associated with adaptive abilities. The authors noted that factors such as parental expectations, the emotional climate and organisation within the home, and enrichment opportunities may be important.

As the construct of adaptive functioning assumes modifiability of skills rather than stability, this is an area where environmental influences may be particularly important (Hauser-Cram et al., 1999). Of interest here is whether there is evidence that the family environment in areas of relationships, personal growth and family system maintenance is related to the adaptive functioning skills in individuals with WS.

Aims of current study

In light of the above, the aims of the current study were threefold: 1) to document the overall level and profile of adaptive functions in WS, both looking at group trends and at individual variability; 2) to investigate the relationship between adaptive functions and the demographic variables of gender, chronological age, and IQ; 3) to investigate the relationship between adaptive functions and the family environment using the Family Environment Scale (FES) questionnaire. The FES assesses three dimensions of the family environment: the quality of family relationships, the family's emphasis on personal growth, and the emphasis on structure, organisation and rules in running the family. We focused on WS individuals with the standard ~1.6Mb genetic deletion to ensure genetic homogeneity of our sample (Porter et al., 2012).

Hypotheses

In relation to the first aim of the study, while this was the first paper to use the Vineland Adaptive Behavior Scales, Second Edition (Vineland II; Sparrow, Cicchetti, & Balla, 2005) spanning across children and adults with WS, we expected (based on previous research) that the overall level of adaptive functioning would be low and, on average, within the severity range of a mild to moderate disability. We predicted wide variability in overall levels of adaptive functioning and in strengths and weaknesses at the domain and subdomain levels, consistent with the clinical variability observed in WS (Jarrold et al., 1998; Pezzini et al., 1999; Porter & Coltheart, 2005). Moreover, we expected Socialisation to be a relative strength for individuals with WS, particularly at the subdomain level of Coping Skills, which reflects how an individual adapts in social situations (e.g., shows sensitivity to others, is polite, has a high degree of empathy). We did not expect the Socialization subdomain of Interpersonal Relationships to be a strength, particularly given anecdotal reports that people with WS have difficulty interacting with and maintaining friendships with same age peers (Dimitropoulos et al., 2009). In line with findings that higher levels of problem behaviours have been associated with lower adaptive functioning (Dimitropoulos et al., 2009; Di Nuovo & Buono, 2011; Phillips, 2008), we expected higher levels of maladaptive behaviours to be negatively associated with adaptive functioning skills.

In relation to the second aim, we hypothesised that there would not be any gender differences in adaptive functions, in line with previous studies (Fisch et al., 2007, Fisch et al., 2010; Fu, 2012; Greer et al., 1997). As found in previous studies on WS, we expected overall adaptive functioning (ABC score), and Communication skills, in particular, to plateau with age. Regarding maladaptive behaviours, we expected age related effects with possibly higher levels of externalising behaviour for younger individuals and higher levels of internalising behaviours for older individuals in-line with previous research (Dodd & Porter; 2009; Porter et al., 2009). Finally, given the wide array of correlations reported between IQ and adaptive functioning, and because this was the first study to use the updated Vineland II scales combined with the Woodcock Johnson Test of Cognitive Abilities – 3rd Edition (WJ III COG; Woodcock, McGrew, & Mather, 2001) in WS, no specific hypotheses were generated.

Regarding the third aim, even though this was an exploratory study into the relationship between adaptive functioning and family environment factors, we expected to find some relationships.

Method

Participants

Forty individuals with WS (21 males and 19 females) were recruited through the Williams Syndrome Associations of NSW, Victoria or South Australia for participation in the study. WS diagnoses were confirmed genetically with a positive FISH test showing the elastin gene deletion (Ewart et al., 1993). Full genotyping was also undertaken (see Porter et al., 2012 for details), which identified ten individuals with an atypical large deletion of ~1.8Mb. These participants were excluded from the study,[3] leaving a genetically homogenous sample of 30 individuals (19 males and 11 females) with the typical ~1.6Mb deletion. Demographic information on the final sample is shown in Table 1, including chronological age (CA), mental age (MA), IQ and Socio-Economic Status (SES). MA and IQ were assessed on the Woodcock-Johnson III Tests of Cognitive Ability, Australian Adaptation or WJ III COG (Woodcock et al., 2001)[4]. Area of residency for each family (using postal codes) was used as a measure of each family's SES based on the Index of Relative Socio-economic Disadvantage (IRSD) created by the Australian Bureau of Statistics (ABS, 2006) and available online at the ABS website. Socio-Economic Index for Areas (SEIFA) percentile score was used, with a lower percentile number reflecting relatively greater disadvantage (lower SES). Table 1 shows the participants are a typical WS cohort, with mild to moderate level IQ on average and SES is representative of the Australian population, covering the range from low to high SES.

Table 1

Demographic Information for the WS Study Cohort

	Mean (SD) [Range]		
	Total Sample (*N* = 30)	Males (*n* = 19)	Females (*n* = 11)
Age	18:4 (8:10) [6:0-39:4]	19:4 (9:3) [6:0-39:4]	17:10 (8:7) [10:2-32:10][a]
IQ (GIA)	61.68 (15.83) [25-92]	61.50 (16.41) [25-92]	62.00 (15.58) [33-89][a]
Mental Age	6:11(1:8) [4:8-10:10]	6:10 (1:8) [4:8-10:10]	7:1 (1:9) [4:11-10:6][a]
SES	61.53 (30.39) [3-100]	64.58 (28.10) [13-100]	56.27 (37.51) [3-97][a]

Note. Age is represented as years:months. For IQ and mental age, *n* = 28.

[3] An analysis indicated that for individuals with the 1.8 Mb gene deletion, their mean level of intellectual functioning (GIA or IQ) was significantly higher than their mean level of adaptive functioning ($t(9) = 2.28$, $p = .05$), whereas for the group with a typical (~1.6Mb) deletion size, there was no significant difference between IQ and adaptive functioning ($p = .23$).

[4] Two participants were unable to complete IQ testing due to personal circumstances. Therefore, mean IQ and mean mental age is for *n* = 28 (18 males and 10 females).

[a]No significant difference between males and females in chronological age [$t(28) = -.247$, $p = .807$]; IQ [$t(26) = -.079$, $p = .938$]; mental age [$t(26) = -.363$, $p = .720$]; or SES [$t(15.69) = .649$, $p = .526$].

Measures

Vineland Adaptive Behavior Scales, Second Edition. Adaptive behaviour was assessed using the Survey Interview Form of the Vineland Adaptive Behavior Scales, Second Edition (Vineland II; Sparrow et al., 2005), an individually administered measure of adaptive functioning with standardised normative data covering birth to 90 years. The Vineland II is a significant revision and update of the VABS and is regarded as having strong psychometric properties (Widaman, 2010). The age range has been expanded compared to the previous edition (VABS) which had normative data up to age 18 years. The Vineland II measures four broad domains of adaptive functions: Communication; Daily Living Skills; Socialisation; and Motor Skills. The Motor Skills domain is intended for individuals aged from birth up to the age of seven years. As participants were not within the age range for the Motor Skills domain, this study will not include Motor Skills data. Each broad domain is comprised of subdomains. Domains and subdomains are described in Table 2. The broad domains combine to provide an overall level of adaptive functioning known as the Adaptive Behavior Composite (ABC). The Motor Skills domain does not contribute towards the overall ABC score for individuals 7 years and older. Three subscales (Internalizing, Externalizing and Other) make up an optional Maladaptive Behavior Index, a measure of problem behaviours. Unlike the domain and subdomain scores, high scores on the Maladaptive domain are undesirable. More severe behaviours can be also be evaluated on the Maladaptive Critical Items but these items do not contribute to a subscale or composite.

Table 2

Domains and Subdomains of the Vineland II

Domains	Subdomains
Communication	<u>Receptive</u>: how an individual listens, pays attention and what he/she understands from spoken language

	Expressive: what an individual says, how he/she uses words and sentences to gather and provide information
	Written: what an individual understands about how letters make words and ability to read and write
Daily Living Skills	Personal: how an individual eats, dresses and conducts personal hygiene
	Domestic: what household tasks an individual performs
	Community: how the individual uses time, money, telephone, computer, and job skills
Socialisation	Interpersonal Relationships: how the individual interacts with others
	Play and Leisure: how an individual plays and uses leisure time
	Coping Skills: how an individual demonstrates responsibility and sensitivity to others
Adaptive Behavior Composite (ABC)	Composite of Communication, Daily Living Skills, and Socialisation domains
Maladaptive Behavior	Maladaptive Behavior Index: composite of Internalizing, Externalizing and Other types of undesirable behavior that may interfere with adaptive functioning
	Maladaptive Behavior Critical Items: more severe maladaptive behaviors

Note. Adapted from *Vineland Adaptive Behavior Scales, Second Edition (Vineland II), Survey Forms Manual* (p. 3), by S.S. Sparrow, D.V. Cicchetti and D.A. Balla, 2005, Circle Pines, MN: American Guidance Service, Inc. Copyright © 2006 NCS Pearson, Inc. Adapted with permission. All rights reserved.

The Survey Interview Form of the Vineland II has very good to excellent levels of reliability. Internal consistency reliabilities for the domains and ABC scores are very high, with most reliabilities being in the upper .80s to low .90s. Subdomain reliability estimates are moderate to high, with around 75% being higher than .75 or more. The average test-retest reliability coefficients across domains range from .88 to .92 and coefficients of subdomains are very high (most values exceeded .85). The Vineland II manual provides theoretical and empirical evidence supporting the validity of the instrument (Sparrow et al., 2005).

The Woodcock Johnson Tests of Cognitive Abilities, Australian Adaptation (WJ III COG). Participants were administered the Woodcock Johnson Test of Cognitive Abilities – 3rd Edition, Australian Adaptation (WJ III COG; Woodcock et al., 2001). Some WJ III subtests are appropriate for individuals as young as 24 months and all tests can be used with individuals aged from 5 to 95 years (Schrank, Flanagan, Woodcock, & Mascolo, 2002). The battery is based on a well-validated theory of cognitive abilities (the Cattell-Horn-Carroll or CHC theory). CHC theory is a combination of the G*f*-*Gc* theory of *fluid* (G*f*) and *crystallized* (G*c*) intellectual abilities purported by Cattell and Horn and Carroll's three-stratum theory of the structure of cognitive abilities (McGrew & Woodcock, 2001). The CHC theory provides support for the design and interpretation of the WJ III COG at three levels. Firstly, the battery consists of 20 cognitive ability tests which measures many of the specific (or narrow) abilities of Stratum I. The test provides measurement of several G*f*-G*c* factors or broad cognitive abilities (Stratum II): Comprehension-Knowledge (*Gc)*, Long-Term Retrieval (*Glr)*, Visual-Spatial Thinking (*Gv*), Auditory Processing (*Ga*), Fluid Reasoning (*Gf*), Processing Speed (G*s*) and Short-Term Memory (*Gsm*). Each factor is derived from two qualitatively different narrow (Stratum I) abilities: *Gc* (Tests 1 and 11), *Glr* (Tests 2 and 12*)*, *Gv* (Tests 3 and 13), *Ga* (Tests 4 and 14), *Gf* (Tests 5 and 15), *Gs* (Tests 6 and 16) and *Gsm* (Tests 7 and 17). The Stratum II level factors provide information for determining cognitive strengths and weaknesses. Finally, an overall (Stratum III) measure of General Intellectual Ability (GIA) or *g* is available (similar to Full Scale IQ. A brief description of the ten subtests which make up the Standard Battery and the ten subtests of the Extended Battery is provided in the Appendix, for further details see the examiner's manual of WJ III COG (Mather & Woodcock, 2001).

The WJ III COG technical manual (McGrew & Woodcock, 2001) provides comprehensive information about reliability and validity. The majority of reliabilities for individual subtests are at the desired level of .80 or higher while the median reliabilities for each of the cluster scores are .90 or higher. The GIA scores have the highest reliabilities of all scores, with the median reliability of the GIA-Std at .97 and GIA-Ext at .98. Validity is based on several sources of evidence including mapping of test content to CHC theory, empirical evidence from the normative sample and confirmatory factor analysis (Schrank et al., 2002).

Family Environment Scale. The Family Environment Scale (FES; Moos & Moos, 1994) is a 90-item true-false measure of the social environment of families. There are three forms of the FES which can be used: the Real Form (Form R), the Ideal Form (Form I), and the Expectations Form (Form E). The Real Form, which was used in this study, measures people's perceptions of their current family environment, while the Ideal Form measure people's preferences about an ideal family environment and the Expectations Form measures expectations about family settings. According to the FES Manual – Third Edition (Moos & Moos, 1994), the Real Form of the FES has been used by clinicians and researchers to describe and compare family climates, understand the impact of the family on children and adolescents with behavioural, emotional or developmental disabilities and understand the role of the family in coping with life transitions and crises. Ten subscales assessing various aspects of the family psychosocial environment create three underlying sets of dimensions: relationships, personal growth (or goal orientation) and system maintenance. See Table 3 for a description of the subscales of the FES.

On the FES, an individual's raw score is converted to a standard score (T-score) with a mean of 50 and standard deviation of 10. Higher scores indicate more of the stated construct and a subscale T-score $\geq$ 60 is considered elevated. Normative data for the FES was obtained from 1432 'normal' families and 788 'distressed' families. According to Moos and Moos (1994), the Family Environment Scale has acceptable internal consistency, with the Chronbach's Alpha coefficient ranging from .61 to .78 across the ten subscales. Test-retest reliability at two months ranged from .68 to .86 across the subscales and at four months ranged from .54 to .86.

Table 3

FES, Broad Dimensions, Subscales and Descriptions

Subscale	Description
Relationship Dimension	
Cohesion	the degree of commitment, help, and support family members provide for one another
Expressiveness	the extent to which family members are encouraged to express their feelings directly
Conflict	the amount of openly expressed anger and conflict among family members
Personal Growth Dimension	
Independence	the extent to which family members are assertive, are self-sufficient, and make their own decisions
Achievement Orientation	how much activities (such as school and work) are cast into an achievement-oriented or competitive framework
Intellectual-Cultural Orientation	the level of interest in political, intellectual and cultural activities
Active-Recreational activities Orientation	the amount of participation in social and recreational
Moral-Religious Emphasis	the emphasis on ethical and religious issues and values
System Maintenance Dimension	
Organization	the degree of importance of clear organization and structure in planning family activities and responsibilities
Control life	how much set rules and procedures are used to run family

Note. From *Family Environment Scale Manual Third Edition* (p. 1), by R.H Moos and B.S. Moos, 1994, Pal Alto, CA: Consulting Psychologists Press. Copyright (1994) by Mind Garden Inc. Reprinted with permission.

Procedure

Administration. The Vineland II survey interview form was administered via a semi-structured interview with either a parent or primary caregiver according to standardised instructions in the Vineland II Survey Forms Manual (Sparrow et al., 2005). The interview was either completed face-to-face on the same day as the cognitive testing or completed afterwards via telephone. The WJ III COG was administered individually to participants using the standardised administration procedure outlined in the examiner's manual (Mather & Woodcock, 2001). Subtests 4, 7, 8, 9, 14 and 17 are typically presented by audio recording, but can be presented orally if subjects have difficulty paying attention or dislike wearing headphones (Schrank et al., 2002). To maintain participant's attention, all subtests were presented orally, apart from subtest 14 for which the audio recording is essential because this test measures speech-sound discrimination. The audio recording provides simultaneous presentation of oral language with increasing louder background noise. Both the Vineland II and the WJ III COG were administered by the first author of this paper; a trained intern clinical neuropsychologist. Administration and scoring was overseen by the second author, a fully registered clinical neuropsychologist. The FES was administered to the family member who had completed the Vineland II to assess their perceptions of their family environment. The FES questionnaire was completed by 27 families (three families did not complete the scale due to personal reasons).

Scoring. All tests were hand scored using standardised instructions outlined in the administration manuals (Sparrow et al., 2005; Mather & Woodcock, 2001, Moos & Moos, 1994) and Vineland II and WJ III COG scores were then double checked via computer scoring programs. The Vineland II was scored using the Vineland II ASSIST (2008) Scoring and Reporting computer program. Computer scoring of the WJ III COG was completed using the WJ III Normative Update (NU) Compuscore and Profiles Program – Australian Adaptation, Version 1.0 (Schrank & Woodcock, 2009). The FES was hand scored using the scoring template provided by the test publisher (Moos & Moos, 1994).

Results

Statistical results were obtained from IBM SPSS Statistics Standard Grad Pack 21 computing software. The level of significance was adjusted for multiple comparisons by using family wise Bonferroni corrections throughout.

The results are presented at three different levels. First the overall adaptive functioning composite (ABC) results for the group are provided. Then the results for the three domains will be presented, followed by the subdomain results. Within the domain and subdomain level, the group averages will be explored and also individual differences. Following the results of adaptive functioning, results of maladaptive behaviours will be presented.

Profile of Adaptive Functioning: Adaptive Behavior Composite (ABC)

Descriptive statistics for the Adaptive Behavior Composite (ABC), domain and subdomain standard scores on the Vineland II are presented in Table 4. ABC and domain scores in Table 4 represent standard scores with a mean of 100 and a standard deviation of 15. The Vineland II manual (Sparrow et al., 2005) defines adaptive functioning as impaired for scores that are two standard deviations below the mean (scores 70 and below). According to this criteria, 93% of the sample (n = 28) were functioning at an impaired level, while the remaining 7% (n = 2) were functioning within the normal (unimpaired) range[5]. Further breakdown of those individuals in the impaired range showed 21 participants (75%) at a Mildly Impaired level (ABC score 50 to 70), three participants (11%) were at a Moderately Impaired level (ABC score 35-49) and four participants (14%) were at a Severely/Profoundly Impaired level (ABC score < 35) [6].

[5] Of the two individuals within the unimpaired range, one had a GIA (IQ) score of 65. GIA was not available for the other participant.

[6] Traditional IQ ranges, based on the American Psychiatric Association's Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition, Text Revised (APA, 2000; DSM-IV-TR) criteria and used by Sparrow et al. (2005) to distinguish levels of severity for individuals with intellectual impairment, were used to describe Low levels of adaptive and intellectual functioning.

Profile of Adaptive Functioning: Domains

As shown in Table 4, for the overall sample, the lowest domain score, on average, was Communication, followed by Daily Living Skills. The highest score was Socialisation. A one-way repeated measures analysis of variance (ANOVA) was conducted to compare performance across the domains of Communication, Daily Living Skills and Socialisation. There was a significant main effect of domain, Wilks's lambda = .63, $F(2, 28) = 8.125$, $p = .002$, $\eta_p^2 = .367$. Follow-up pairwise post-hoc comparisons revealed that Socialisation was a significant strength, on average, compared to Communication ($p = .009$). Neither the differences between Communication and Daily Living Skills nor between Socialisation and Daily Living Skills reached significance ($p = .546$ and $p = .073$, respectively).

Table 4

Mean ABC, Domain and Subdomain Scores on the Vineland II (N=30)

Domains[a] Subdomains[b]	Mean (*SD*)	Range
Adaptive Behaviour Composite (ABC)	**55.97 (15.11)**	**20 – 84**
Communication	**53.83 (19.51)**	**21 – 82**
Receptive	7.67 (2.48)	1 – 12
Expressive	7.83 (2.07)	2 – 10
Written	8.03 (2.54)	2 – 14
Daily Living Skills	**57.40 (14.01)**	**21 – 93**
Personal	7.63 (2.57)	1 – 14
Domestic	7.13 (2.85)	1 – 16

Community	7.40 (3.06)	1 – 13
Socialization	**60.77 (14.44)**	**20 – 83**
Interpersonal Relationships	7.87 (2.36)	1 – 11
Play & Leisure Time	8.23 (1.96)	3 – 12
Coping Skills	9.23 (2.11)	4 – 14
Maladaptive Behavior Index[c]	**17.60 (2.54)**	**10 – 24**
Internalizing	18.63 (2.40)	13 – 24
Externalizing	16.47 (2.30)	12 – 22

Note. [a]ABC and domain standard scores (in bold) represent standard scores with a mean of 100 (*SD* = 15), range 20-160. Scores ≤ 70 are impaired. The ABC score is obtained from the sum of domain standard scores. For each domain, normative tables provide the domain score from the sum of the relevant subdomain *v*-scale scores. [b.]Subdomain *v*-scale scores with a mean of 15 (*SD* = 3), range 1-24. Scores ≤ 9 are impaired. [c]Maladaptive Behavior Index is obtained from the sum of Internalising, Externalising and Other raw scores. Maladaptive Behavior Index, Internalising and Externalising scores are *v*-scale scores, but in contrast to subdomain scores high scores are undesirable. For Maladaptive domain, three categories are used to describe performance: Average level = *v*-scale score below 18, Elevated level = *v*-scale range of 18-20, Clinically Significant level = *v*-scale score range 21-24.

Intra-individual Comparisons

An individual's domain strengths and weaknesses, relative to their own level of ability, were calculated by the Vineland II scoring program. This method of *intra-individual comparison* compares an individual's score to his or her own unique profile by comparing each domain score with their own median domain standard score (Sattler, 2001). In line with Sattler, the Vineland II scoring program considers a difference of 10 points or more, either a significant personal strength or a personal weakness. Using this criteria, Figure 1 indicates the percentage of individuals identified as having either a personal strength or weakness (or neither a strength nor weakness) for each domain.

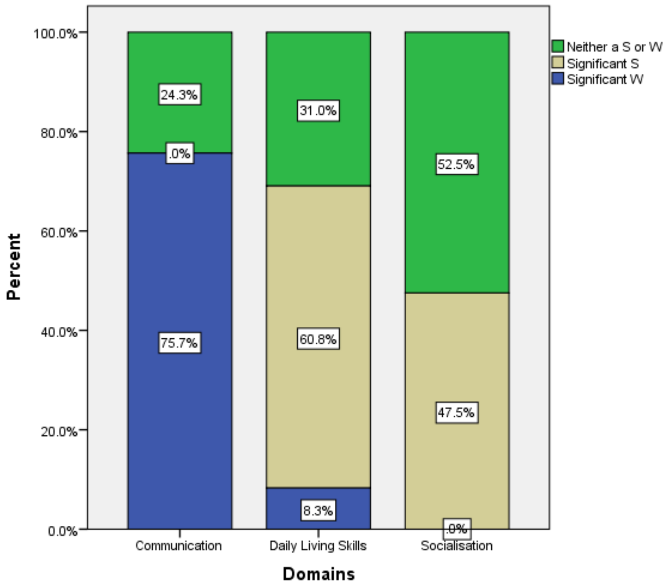

Figure 1. **Percentage of individuals who were found to have either a personal strength (S) or personal weakness (W), or neither a strength nor a weakness, for each**

Domain of Adaptive Functioning. Figure 1 shows great variability in Domain strengths and weaknesses. Overall, almost half of the individuals (47%) showed a personal strength in Socialisation skills, while the other half (53%) showed neither a strength nor a weakness in Socialisation skills. No individual showed a significant weakness in Socialisation. Therefore, the group mean pattern of Socialisation being a relative strength is not evident for half of the individuals in this study. For Daily Living skills, nearly one third of the group (31%) showed no strength or weakness and the majority (61%) showed a significant personal strength in their Daily Living Skills, which was not identified at the group level. Communication skills were a significant weakness for the majority of individuals (72%) in line with group findings. No individuals had a personal strength in Communication skills.

Profile of Adaptive Functioning: Subdomains

Group Level. Subdomain scores on the Vineland II are shown in Table 4. The subdomain scores are standardised scores (*v*-scale scores), with a mean of 15 and standard deviation of 3 (scores ≤ 9 are impaired). One-way repeated measures ANOVAs were used to compare scores at the Subdomain level. There was no significant difference between the three Communication subdomains (Receptive, Expressive, Written), $F(2, 28) = 0.420$, $p = .661$ or the Daily Living Skills subdomains (Personal, Domestic, Community), $F(2, 28) = 0.653$, $p = .528$. There was a significant main effect for the Socialisation subdomain, Wilks's lambda = .57, $F(2, 28) = 10.442$, $p < .0005$, $\eta_p^2 = .43$. Bonferroni adjusted comparisons revealed that within the Socialisation domain, Coping Skills were a significant strength, overall, compared to both Interpersonal Relationships ($p = .001$) and Play and Leisure Time ($p = .005$). There was no difference between Interpersonal Relationships and Play and Leisure Time ($p = .814$).

Intra-individual subdomain comparisons were also computed by the Vineland II scoring program, where the individual's v-scale score was compared to their median v-scale score for that domain and as outlined by the Vineland II manual (Sparrow et al., 2005) a difference of 2 points or greater was considered meaningful. Figure 2 shows the percentage of individuals identified as having a relative subdomain strength or weakness (or neither a strength nor weakness).

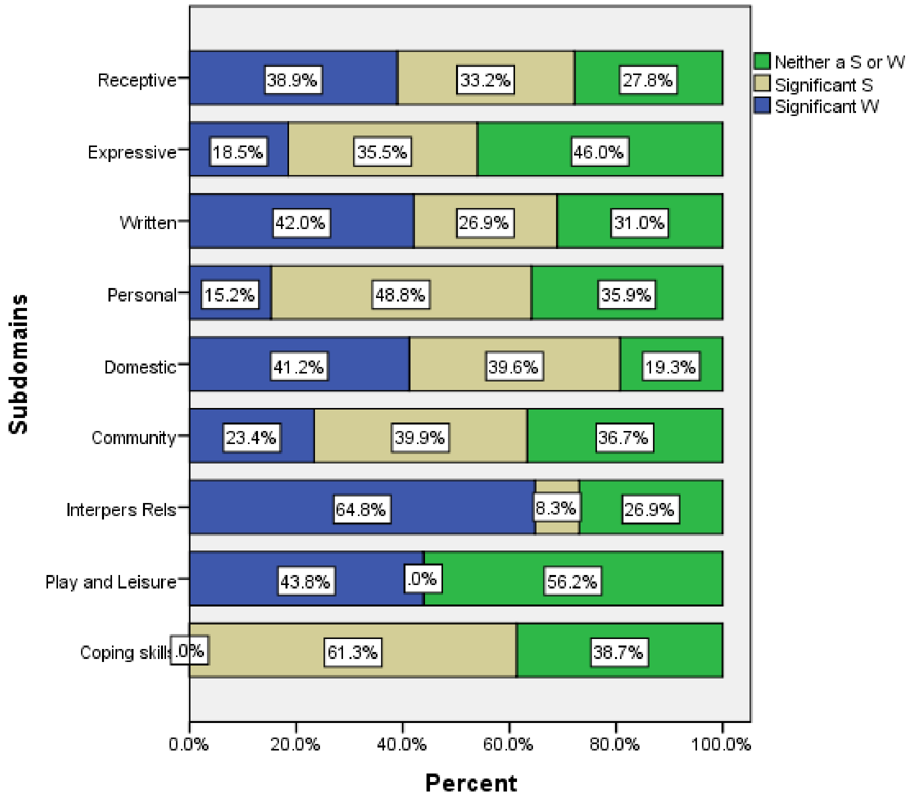

Figure 2. **Percentage of individuals who were found to have either a personal strength (S) or personal weakness (W), or neither a strength nor a weakness, for each Subdomain.**

As shown in Figure 2, vast heterogeneity is evident at the subdomain level. While at the group level, there were no significant differences between the subdomains which comprise the Communication and Daily Living Skills domains, there are different relative strengths and weaknesses at the individual level. Within the Communication domain, relative weaknesses were highest for Written skills (42% of individuals), followed by Receptive skills (39%) and relatively less for Expressive skills (18%). Percentage of personal strengths within the Communication domain were similar: Expressive (35%), Receptive (33%) and Written (27%). Within the Daily Living Skills domain, the greatest relative weakness was for Domestic skills (41%), followed by Community skills (23%), and then Personal skills (15%). Relative strengths were largest for Personal skills (49%), followed by Domestic and Community skills (both 40%). Within the Socialisation domain, the majority of individuals (65%) displayed a relative weakness in Interpersonal Relationships and only a small percentage had a relative strength (8%). For Play and Leisure skills, 44% had a relative weakness and no individual displayed a personal strength. The majority of individuals

(61%) had a relative strength in their Coping Skills which reflects the findings at the group level. No individual had a relative weakness with Coping Skills.

Maladaptive Behaviour Index. Means and standard deviations for the Maladaptive Behaviour Index and for Internalising and Externalising behaviours are shown in Table 4. Forty three percent (n = 13) of participants were found to have an Elevated score on the Maladaptive Behavior Index (more problem behaviours than 84% of same aged individuals) and 10% (n = 3) demonstrated Clinically Significant levels of overall maladaptive behaviour (a score in the extreme 2% of individuals the same age). A paired samples t-test was conducted to evaluate whether there was a significant difference between the level of internalising and externalising behaviours. There was a statistically significant difference, $t(29) = 5.78$, $p < .0005$, with participants showing significantly higher levels of Internalising than Externalising behaviours, on average. The eta squared statistic (.54) indicated a large effect size (Cohen, 1988). A breakdown of the level of these behaviours revealed that 47% (n = 14) of participants had an Elevated level of Internalising behaviours and 20% (n = 6) a Clinically Significant level, while for Externalising behaviours 23% (n = 7) were Elevated and 7% (n = 2) at a Clinically Significant level.

The most commonly reported Internalising problem was for feelings of anxiety (77%), followed by eating difficulties (63%). Eating difficulties referred to problems with either the rate of eating, hoarding food, overeating or refusing to eat. The most common Externalising problem was impulsivity (63%) and failing to inhibit inappropriate comments or questions in public (57%). Other common problem behaviours included difficulty paying attention (80%) and acting overly familiar with strangers (47%). While the Critical items do not form part of the Maladaptive Behavior Index, an examination of the frequency of responses indicated that the most common problem reported was fear of ordinary sounds, objects or situations (70%).

Relationship between Adaptive and Maladaptive Behaviours. Correlations between Internalising, Externalising behaviours and subdomains of adaptive functioning were undertaken to determine whether problem behaviours impacted on particular areas of adaptive abilities. Higher levels of Internalising behaviours were associated with lower Expressive Communication skills ($r = -.534$, $p = .002$) and lower Interpersonal Relationships ($r = -.429$, $p = .018$).

Relationship between Adaptive Functioning and Chronological Age, IQ, and Gender

Relationship between Adaptive Functioning and Gender. An independent samples t-test was conducted to compare the difference in overall level of adaptive functioning (ABC) for males and females. There was no significant difference, $t(28) = 0.511$, $p = .614$. Independent t-tests were also conducted to compare domain and subdomain performance between males and females. There was no significant difference between males and females on the three broad domains of Communication, Daily Living Skills and Socialisation (all p values > .309) or their subdomains (all p values > .056). There was no significant difference between males and females on the Maladaptive Behavior Index [$t(28) = -1.43$, $p = .165$] nor for levels of Internalising [$t(28) = -1.28$, $p = .210$] or Externalising [$t(28) = -1.31$, $p = .200$] behaviour.

Relationship between Adaptive Functioning and Chronological Age. Pearson correlations were used to investigate the relationship between chronological age and adaptive functioning. A significant negative correlation was obtained between chronological age and ABC score [$r = -.777$, $p < .0005$] indicating that the overall level of adaptive functioning was lower for older individuals. Significant negative relationships were also evident for the domains of Communication [$r = -.854$, $p = .002$], Daily Living Skills [r = -.542, $p = .002$] and Socialisation [$r = -.670$, p < .0005].

To further investigate age related differences, the sample was divided into two age groups (Howlin et al., 2010), with a younger age group consisting of individuals still attending school, aged 6-16 years ($n = 15$) and an older group who had finished formal schooling and were aged 18 years or above ($n = 15$) were formed. Table 5 shows descriptive statistics separately for the two age groups, including chronological age, IQ, and mental age. The younger group was made up of 7 males and 8 females, while the older group had 12 males and 3 females. There was no difference between the two age groups on level of intellectual functioning (GIA) [$t(26) = -.643$, $p = .526$] or mental age [$t(26) = -1.836$, $p = .078$].

Table 5

Descriptives of Younger and Older Groups

	n	FSIQ M (*SD*) [Range]	CA[a] M (*SD*) [Range]	Mental Age M (*SD*) [Range]
Younger Group	15	59.87 (14.62) [33 - 92]	11y5m (2y11m) [6y0m - 15y9m]	6y5m (1y3m) [4y8m - 9y0m]
Older Group	15	63.77 (17.48)[b] [25 - 89]	25y3m (7y2m) [18y4m - 39y4m]	7y6m (1y11m) [5y0m - 10y10m]

Note. [a]CA = Chronological Age, [b]IQ for Older Group is based on n = 13

There was a significant difference in overall level of adaptive functioning (ABC) across the two age groups, t (14.99) = 3.06, p = .008, (Cohen's d = 1.12), with the younger age group having a significantly higher mean ABC score (M = 63.40, SD = 3.48) than the older group (M = 48.53, SD = 18.50). Figure 3 shows the patterns of means for the younger and older age groups, across the domains of Communication, Daily Living Skills and Socialisation. An ANOVA with Age Group as the between groups factor (2 levels: younger, older) and Domain as within groups factor (3 levels: Communication, Daily Living Skills, Socialisation) was conducted to compare differences in domain scores for the two age groups. A significant interaction effect of Domain by Age Group was found. Mauchly's test indicated that the assumption of sphericity had not been met, $\chi^2(2) = .787$, $p = .04$. With Greenhouse-Geisser adjustment ($\varepsilon = .82$), [$F(1.65, 46.15) = 12.47$, $p < .0005$, $\eta_p^2 = .308$.

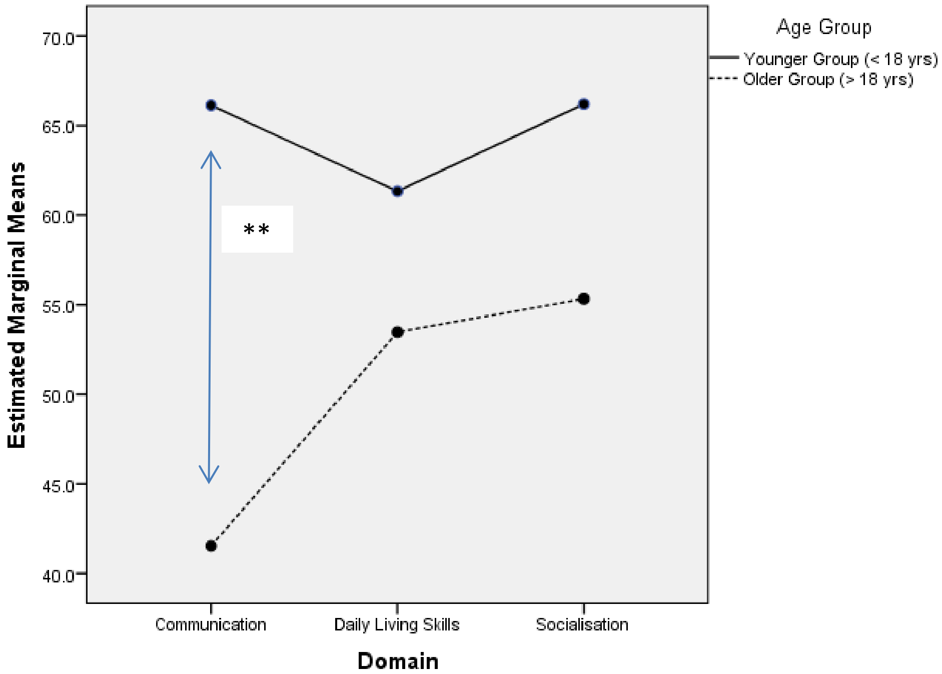

Figure 3. **Mean domain scores for younger and older age groups. Figure 3 shows significant difference between groups on Communication (** *p* < .005).**

Follow-up pairwise comparisons were made to examine the differences between Vineland II domains for each age group. For the younger age group, there were no significant differences between domain scores (Communication vs Daily Living Skills, $p = .122$; Communication vs Socialisation, $p = .979$; the difference between Daily Living Skills and Socialisation approached significance, $p = .022$). For the older group, both Daily Living and Socialisation skills were significantly higher than Communication skills ($p < .0005$ for both comparisons). There was no difference between Daily Living and Socialisation skills ($p = .359$). Pairwise comparisons found no significant difference between the older and younger groups on their level of Daily Living Skills ($p = .126$), while the difference between Socialisation abilities approached significance ($p = .037$). There was a significant difference between the two age groups on the Communication domain, with the younger group's score ($M = 66.13$, $SD = 3.93$) significantly higher than the older group ($M = 41.53$, $SD = 3.93$), $p < .0005$.

At the subdomain level, there was no significant interaction between age group (younger and older) and the three Communication subdomains [$F(2,56) = 0.106$, p =

.900], nor the three Daily Living Skills subdomains [$F(2,56) = .763$, $p = .471$]. There was a significant interaction between age group and the Socialisation subdomains. Univariate results are presented as Mauchly's test indicated that the assumption of sphericity was met, Mauchly's W = .969, $\chi^2(2) = .839$, $p = .658$, Greenhouse-Geisser ($\varepsilon = .970$) and Huynh-Feldt ($\varepsilon = 1.0$) [$F(2,56) = 9.61$, $p < .0005$, $\eta_p^2 = .255$]. Follow-up pairwise comparisons revealed the only significant difference between the groups was for Interpersonal Relationship skills, with the younger group ($M = 8.87$, $SD = .559$) having significantly higher skills than the older group ($M = 6.87$, $SD = .559$), $p = .017$. The Interpersonal Relationship skills for the older age group were also significantly lower than their Play and Leisure (p < .01) and Coping Skills ($p < .0005$).

Maladaptive Behaviour. There was no significant difference between the younger and older age groups on overall level of Maladaptive Behaviours, [$t(28) = -.425$, $p = .674$] or Externalising behaviours [$t(28) = -.788$, $p = .437$]. There was a trend towards significance for the older group displaying higher levels of Internalising behaviours [$t(28) = -2.18$, $p = .038$, Cohen's $d = 0.797$].

Relationship between Adaptive Functioning and IQ

Both IQ (as measured by GIA standard score of WJ III COG) and ABC scores have been standardised on the same scale ($M = 100$ and $SD = 15$), so are directly comparable. As can be seen from Table 1 and Table 4, the spread of scores for intellectual functioning (GIA) and adaptive behaviour (ABC) are similar, ranging from 25 to 92 and 20 to 84, respectively. A paired samples t-test found no significant difference between mean GIA and ABC scores [$t(27) = 1.217$, $p = .234$]. The proportion of the sample that showed a significant difference between their GIA score and adaptive behaviour ABC score was calculated with a significant difference defined as at least one standard deviation (15 points) between the GIA and ABC scores.[7] For

[7] It is important when evaluating the difference between two scores to consider reliability and errors of measurement (Harvill, 1991). A difference equal to or greater than two Standard Errors of Measurement (SEMs) is often used to represent a real difference or change between scores. The SEM for the WJ III COG varies across age ranges with the highest value being 2.60 and for the Vineland II the highest SEM is 4.23. Therefore, using one standard deviation of 15 points is a conservative measure of

the 28 participants who had completed both adaptive and cognitive testing, 29% (*n* = 8) had an intellectual ability score (GIA) significantly higher than their adaptive functioning (ABC) score (GIA > ABC of ≥ 15 points), while 25% (*n* = 7) had a significantly higher ABC score than GIA (ABC > GIA of ≥ 15 points).

This pattern was also considered for the younger (*n* = 15) and older (*n* = 13) groups. Both groups showed a similar percentage with their ABC score higher than their GIA score [27% (*n* = 4) for the younger group and 23% (*n* = 3) for the older group]. However the older group had a larger number of participants with their GIA score significantly higher than their ABC score [54% (*n* = 7) for the older group and 7% (*n* = 1) for the younger].

Correlations between General Intellectual Ability (GIA) and cognitive factors from the WJ III COG and overall Adaptive Functioning (ABC) and adaptive functioning domain scores (Communication, Daily Living Skills and Socialisation) on the Vineland II were examined. Due to the number of comparisons being made, the significance level was set at $p < .01$ to control for possibility of Type 1 error. Pearson correlations indicated no significant relationship between GIA and ABC scores ($r = -.06$, $p = .757$) nor between GIA and the domains of Communication ($r = -.138$, $p = .483$), Daily Living Skills ($r = .139$, $p = .481$), Socialisation ($r = -.050$, $p = .800$) or Maladaptive Behaviour ($r = -.087$, $p = .661$), Internalising ($r = -.124$, $p = .529$) or Externalising ($r = .064$, $p = .745$).

There was also no evidence of a relationship between GIA and domain scores on the Vineland II and specific cognitive abilities or CHC factors (Comprehension-Knowledge, Long-Term Retrieval, Visual-Spatial Thinking, Auditory Processing, Fluid Reasoning, Processing Speed, Short-Term Memory) on the WJ III COG (all *p* values > .05).

Family Environment and Adaptive Functioning

Means and standard deviations for the ten subscales of the FES questionnaire are reported in Table 6. Scores are T-scores with a mean of 50 and standard deviation of

---

difference between scores as this is greater than two SEM’s for both measures (Sattler, 2001; Sparrow et al., 2005)

10. A t-score ≥ 60 is considered an elevated score and represents that a family reports more of the stated construct. As shown in Table 6, the highest mean score was on the Cohesion subscale, indicating that the families provide high levels of commitment, help, and support for one another. The lowest mean score was on the Conflict subscale, indicating relatively low levels of openly expressed anger and conflict amongst family members. Partial correlations were used to explore the relationship between an individual's family environment (as measured by the FES), their level of overall adaptive functioning (ABC) and Vineland II domain scores. A *p*-value of .05 was used to indicate significance. While using this approach for multiple comparisons can result in Type One Errors (rejecting the null hypothesis when it is in fact true), due to the exploratory nature of the study and the small sample size, it was decided that this was preferable to making a Type Two Error (failing to reject a null hypothesis when it is false; Rothman, 1990). The significant correlations identified between ABC and domain scores and FES subscales, are presented in Table 7.

Table 6

Family Environment Scale (FES) Questionnaire Subscale Means (and Standard Deviations)

Subscale	Mean (sd) *n = 27*	Range
Cohesion	59.44 (4.85)	52 - 65
Expressiveness	54.04 (7.77)	40 - 71
Conflict	42.96 (7.39)	33 - 65
Independence	46.78 (8.97)	30 - 69
Achievement Orientation	44.96 (9.36)	33 - 59
Intellectual Cultural Orientation	51.19 (12.10)	30 - 69

Active Recreational Orientation	53.63 (12.57)	33 - 69
Moral Religious Emphasis	47.00 (8.86)	32 - 66
Organisation	52.04 (11.91)	32 - 69
Control	52.59 (10.22)	32 - 70

Note: Subscale standard scores are T-scores with a mean of 50 and standard deviation of 10. An elevated T-score ≥ 60, suggests a family reports more of that construct when compared to families which make up the normative data for FES.

As can be seen in Table 7, the Communication domain was not correlated with any of the measured aspects of the Family Environment. Families who rated higher on Intellectual/Cultural outlook and Active Recreation were associated with higher outcomes on their family member with WS's overall adaptive functioning (ABC), Daily Living Skills and Socialization skills. A high level of Control in families was significantly associated with higher Daily Living Skills. For maladaptive behaviours, the only significant correlation was found with externalising behaviour, whereby higher levels of externalising behaviours were associated with low Independence (with regard to self-sufficiency and making their own decisions).

Pearson correlations were conducted to explore the relationships between FES ratings and intellectual functioning (GIA) and FES ratings and the family's level of socio-economic status (SES). No significant correlations were obtained ($p > .1$ for all correlations).

Table 7

Significant Correlations between FES and Vineland II Domains

	Family Environment Subscales			
Domain	Intellectual/ Cultural	Active/ Recreation	Independence	Control
ABC	.450*	.388*		
Communication				
Daily Living Skills	.525**	.512**		.427*
Socialisation	.384*	.389*		
Maladaptive				
Externalising			-.389*	

Note. $p < .05$* $p < .01$**

Discussion

Adaptive functioning was investigated in a group of 30 WS individuals with the typical ~1.6Mb deletion. Group patterns of adaptive functioning were investigated, as well as individual profiles of strength and weakness. As with previous studies on WS, the current study found extensive variability in adaptive functions, at the global level, as well as the domain and subdomain levels. Moreover, group trends were not always representative of patterns at a case-by-case level. Despite genetic homogeneity, the sample comprised of males and females and included children and adults with variable IQs, allowing the authors to investigate the relationship between adaptive functions and gender, chronological age and IQ. Unlike intellectual abilities, which are said to be highly genetic and heritable (Plomin & Thompson, 1993) and to remain stable over time (Porter & Dodd, 2011; Sattler, 2001), adaptive functions are said to be more modifiable by the environment (Sattler, 1992). The current study, therefore, explored whether environmental factors, specifically characteristics of the family environment were related to one’s level of adaptive functioning. Ultimately, the authors were interested in seeing whether demographic or environmental factors could, at least partially, account for some of the variability in adaptive functions observed in individuals with WS. While adaptive functions were not significantly related to gender or IQ, there were relationships between adaptive functions and chronological age. There were also relationships with family environment characteristics. Findings are discussed in more detail below, along with clinical implications.

Adaptive Profile in WS using the Vineland II

As hypothesised, the majority of individuals in the current study demonstrated an impaired level of adaptive functioning, which is consistent with previous studies that have reported deficits in adaptive functioning in WS (e.g., see Fu, 2012; Howlin et al., 1998). The average level of both intellectual functioning and adaptive functioning was within the range of a Mild to Moderate Intellectual Disability, but there was evidence of individual variability in both intellectual and adaptive functions. Intellectual functions ranged from severely impaired (five standard deviations below the population mean) to average and adaptive functions ranged from severely impaired to low average. Of note then, some individuals were in the normal range for intellect and/or adaptive functioning.

At a group level, individuals with WS displayed relative strengths in their Socialisation skills, followed by their Daily Living Skills and were lowest on Communication Skills. Socialisation skills were significantly higher than Communication skills, on average. A relative strength in Socialisation skills was consistent with our hypothesis and the results of previous studies of adaptive function (Howlin et al., 1998, Mervis et al., 2001) and is also consistent with the social and behavioural phenotype of WS. Fu (2012) did not replicate this result and suggested that this may have been due to the wide age range used in their study. However, the current study used a similarly wide age range, so it would appear that Fu's second suggestion - that their findings may have been due to the adaptive behaviour scale used – may better account for their results. Fu used the SIB-R, which combines social interaction and communication skills into the one cluster score, whereas the Vineland scales (including the Vineland II) treat these as two distinct areas of functioning. Given our findings that these two skills dissociate in WS, the Vineland II would appear to be a more appropriate tool than the SIB-R for individuals with WS.

At a group level, there was no significant difference between Daily Living Skills and either Socialisation or Communication. This is different to the results of previous studies, which have found Daily Living Skills to be a significant weakness for individuals with WS (Dimitropoulos et al., 2009, Fisch et al., 2007, Greer et al., 1997; Mervis et al., 2001), although these studies used younger groups comprised of children or children and adolescents, whereas the current study used a wider age range including both children and adults, this does not seem to fully account for the different findings.

At the subdomain level, the only significant difference was found within the Socialisation domain, where Coping Skills were found to be a relative strength, supporting our hypothesis. Coping Skills assesses whether an individual demonstrates responsibility and sensitivity to others. This is consistent with the personality characteristics of WS as being empathetic and attuned to the emotional state of others (Mervis & Klein-Tasman, 2000).

Individual Variability in Adaptive Functions

As has been noted by a number of researchers (e.g., Howlin et al. 2010; Jarrold et al., 1998; Porter & Coltheart, 2005) group data may not reflect the variability in functioning of individuals within the group. In this study, patterns of behaviour at the group level did not necessarily reflect patterns of individual strength and weakness. Individual results confirmed the group finding of Communication skills being a relative weakness. However, Daily Living Skills were rated as a personal strength for nearly two thirds of individuals, which was not reflected at the group level. Socialisation skills were a relative strength at the group level, but this was only evident for a small percentage of individuals when their profiles were further explored and was found to be neither a personal strength nor weakness for just over half of the study cohort.

At the subdomain level, as hypothesised, the Interpersonal Relationships subdomain, which measures social relating and social communication skills, was a relative weakness for many participants. Due to their outgoing nature, most individuals with WS are happy to initiate conversation with other people, however some difficulties in the social use of language (pragmatic language) reported in WS, including the overuse of stereotyped responses, inappropriate initiation of conversations, and perseverative responding (see review by Brock, 2007; Laws & Bishop, 2004; Mervis & John, 2010) may impact on their social communication skills. This may also reflect the findings that individuals with WS experience difficulties maintaining friendships (Dimitropoulos et al., 2009). Overall, these findings highlight that is it important to consider both group level and individual level data when investigating adaptive functioning in WS, as has been shown with cogntive and other clinical characteristics.

Relationship between Maladaptive and Adaptive Behviaour

Consistent with previous research, which has reported that up to 60 to 70% of individuals with WS have elevated levels of problem behaviours (Einfeld et al., 1997; Greer et al., 1997; Udwin & Yule, 1991), nearly half of the current WS sample was found to have problem behaviours. Also consistent with previous research (Einfeld et al., 1997; Kennedy et al., 2006) our sample displayed significantly higher levels of internalising behaviours than externalising behaviours, with feelings of anxiety the most commonly reported problem. This is highly consistent with the WS phenotype and the high levels of GAD reported in WS (Dodd & Porter, 2009; Dykens, 2003; Leyfer, Woodruff-Borden, Klein-Tasman, Fricke, & Mervis, 2006; Woodruff-Borden, Kistler, Henderson, Crawford, & Mervis, 2010).

We hypothesised that problem behaviours may impact on adaptive skills and this was confirmed with higher internalising behaviours found to be associated with lower

Expressive communication skills and Interpersonal Relationship skills, suggesting that these difficulties may impact on an individual's ability to interact and relate well (social communication) with other people. These results are not consistent with previous findings that reported that higher internalising behaviours were associated with a strength in verbal intellectual skills (Porter et al., 2009). The disparity may perhaps be explained by the different measures used to assess internalising behaviours (Child Behavior Checklist versus Vineland II) across the studies and may reflect differences in the verbal concepts - verbal adaptive measures versus verbal intellect. There was no association found between maladaptive behaviours and chronological age, suggesting that problem behaviours do not necessarily get better or worse with age. However, there was evidence that older individuals may display higher levels of internalising behaviours, consistent with other reports of higher rates of depression and generalised anxiety in adults compared to children with WS (Dodd & Porter, 2009). The lack of a significant result may have been a power issue, with small numbers in each age group. This is reflected by the large effect size for this comparison of 0.797.

Relationship between Adaptive Functioning in WS and Demographic Variables

No Gender Differences in Adaptive Functioning

Males and females with WS displayed similar Vineland II scores across global, domain and subdomain levels. In addition, maladaptive behaviours (internalising/externalising scores) were not different for males and females. Findings of no gender differences are consistent with Fu's (2012) results across a similarly wide age range of WS individuals using the SIB-R scales. Findings also parallel the fact that no gender differences have been observed in cognitive abilities of people with WS in this study and other studies (Greer et al., 1997; but see also Fisch et al., 2010) and parallel findings on the normative population of the Vineland II. The Vineland II manual (Sparrow et al., 2005) reports that mean standard ABC scores and domain and subdomain scores were similar for males and females, which suggests there are no gender differences in adaptive functioning for typically developing individuals.

In comparison to the findings of the current study, Porter et al. (2009) reported that females were significantly more likely to display externalising problems compared to males (measured using the CBCL, Achenbach & Rescorla, 2001). This may reflect genetic heterogeneity in Porter et al.'s cohort. Subsequent genotyping indicated that a large percentage of Porter et al.'s sample had a larger 1.8Mb gene deletion (Porter et al., 2012). Patients with a 1.8Mb deletion have since been found to display more behaviour difficulties and greater executive dysfunction than those with the standard ~1.6Mb deletion (Porter et al., 2012).

Similarly, gender differences in adaptive functions and externalizing behaviours have been identified in Fragile X syndrome, which is an X-linked disorder in which males are more severely affected cognitively, including more severe executive functioning difficulties (Fisch et al., 1999; Fisch et al., 2010; Glaser et al., 2003).

Change in Adaptive Abilities with Age

When the individuals in this study were divided into younger and older age groups, the younger group was found to have significantly higher overall adaptive functions (ABC) and Communication Skills than the older group. While the difference between the higher Socialisation skills of the younger group approached significance, there was no difference between younger and older groups on Daily Living Skills. The pattern of Domain means for the younger group followed the pattern noted by previous studies (Dimitropoulos et al., 2009; Fisch et al., 2007; Greer et al., 1997; Mervis et al., 2001) with Socialisation highest, followed by Communication and Daily Living Skills lowest. The level of these skills was relatively similar, that is, they were not significantly different from one another.

For the older group, a different pattern emerged, with Socialisation skills highest, followed by Daily Living Skills and Communication Skills lowest. Confirming our hypothesis, Socialisation and Daily Living Skills were both significantly stronger than Communication skills. This suggests that as individuals with WS get older, they fail to keep developing their adaptive abilities at the same rate. Further confirming these findings were the negative relationships identified between age and overall adaptive functioning and each domain of adaptive functioning: Communication, Daily Living, and Socialisation skills. While this could be interpreted as a decline in functioning with age, Fisch (2000) points out that adaptive scales are age normed, so a decline in scores relative to chronological age does not mean a loss of ability or regression of skills. Rather, this can reflect a plateauing of abilities, suggesting, for example, that as individuals with WS get older, their level of adaptive functioning falls further behind their same aged peers from the normative population (Fisch et al., 2007).

Interestingly, this apparent plateauing of abilities was not the same across all skill areas, as the Communication skills of older individuals with WS appear to fall substantially further behind in relation to their other adaptive abilities. One possibility for this finding is that the types of skills that are assessed by the Communication domain may be particularly sensitive to the increasing demands placed on individuals with WS as they get older. The Receptive subdomain assesses skills related to listening and attending and following verbal instructions and the Expressive subdomain includes items to assess an individual's ability to initiate and sustain conversation. For example, attention problems would make it difficult to maintain concentration in conversations or during oral presentations for increasingly longer periods of time, as expected of typically developing individuals. Difficulties with sustaining attention and working memory impairments can affect their ability to take in and follow multi-step instructions. Within the Written subdomain, items are related to academic skills (early alphabetic knowledge and reading and writing abilities), which are, in turn, dependent upon cognitive abilities. Therefore, the marked plateauing of Communication skills with increasing chronological age, relative to other adaptive skills may be due to the cognitive phenotype observed in WS.

Given the well-known personality characteristics of WS individuals as friendly, hypersocial, affectionate and empathetic (Riby & Porter, 2010) it could be assumed that Socialisation skills would be a strength and continue to be an area of strength and an advantage across the lifespan for WS. Therefore, it may be surprising to find that the Socailsiation skills for adults with WS were found to be lower than those of younger individuals. The difference is likely due to the increased social demands typically expected with increased age. These are common areas of concern expressed by parents regarding their child with WS. They describe the difficulties their children experience making and maintaining friendships and observe the gap widening between their child and same aged peers as the social demands change with increasing age. The Vineland II has included additional items within the Socialisation domain (compared to the VABS) to assess an individual's ability to understand and use nonverbal communication skills such as recognising social cues, along with items to assess gullibility, social naïveté and their ability to recognise and avoid victimisation (Sparrow et al., 2005). It has been reported that the friendly and approaching nature of individuals with WS can place them at risk in social interactions due to problems with impulsivity and indiscriminate over friendliness, resulting in exploitation (Davies et al., 1997; Dimitropoulos et al., 2009; Dykens & Hodapp, 2007). A lack of development in Socialization skills with age may also reflect the social isolation experienced by adults with an intellectual disability after they leave the structured school setting (Sparrow et al., 2005), especially as many individuals do not obtain regular work. The implication for individuals with WS is, that despite their friendly nature and willingness to engage in social interactions, they may require organised recreational and social opportunities to enable them to meet and socialise with appropriate peers.

While the patterns of relative strengths and weaknesses identified in the sample of WS, for example their greater weaknesses in Communication skill for older compared to younger individuals, can also be consistent with intellectual disability (see Sparrow et al., 2005), the same patterns are not seen across all neurodevelopmental syndromes with an associated intellectual disability (e.g. see Dimitropoulos et al., 2009; Di Nuovo & Buono, 2011; Fisch et al., 2007). Di Nuovo and Buono (2011) concluded in their study which included five of the most frequent genetic syndromes (Down, Williams, Angelman, Prader-Willi, and Fragile-X) there are similarities and differences in their adaptive profiles. Therefore, it would appear that there are some aspects of the adaptive strengths and weaknesses identified which are more specific to WS.

Unconfirmed Relationship between Intellectual and Adaptive Functioning

It is of interest to know whether the same individuals who are functioning either within the Adequate or Average level for adaptive functioning also function at this level intellectually. It would appear that this is not necessarily the case in WS. The difference between adaptive functioning and intellectual ability was assessed for each participant and more than half of the individuals had a statistically significant difference between these two abilities, either with their adaptive skills significantly higher than their intellectual skills or, conversely, their intellectual skills significantly higher than their adaptive abilities. When the age group of the participants was considered, it was found that those with an IQ score higher than their adaptive functioning score were predominantly from the adult group. This suggests that as individuals with WS get older, they are failing to make the appropriate gains over time in regards to their adaptive functioning, and as individuals with WS get older, there is a larger discrepancy between their level of intellectual and the level of adaptive functioning. A possible contributing factor to this may be that the individuals in the older age group had left the formal school environment. Perhaps the structure of the school setting, where an individualised education plan would be developed for students with an intellectual impairment, provides more challenges and opportunities to develop skills. This highlights the importance of ongoing assistance for individuals with WS beyond the formal schooling years, for example, by taking part in transition to work programs.

Similarly, there was no significant relationship between intellectual ability (IQ) or more specific cognitive abilities (measured by the WJ III COG) and overall adaptive functioning or adaptive domain scores. This is consistent with the results of some previous studies (e.g., Di Nuovo & Buono, 2011; Greer et al., 1997) but inconsistent with other studies (e.g., Fu, 2012; Mervis et al., 2001). The varying results in the literature may reflect differences in the scales used to assess these constructs and in demographic variables such as chronological age.

The Influence of the Family Environment

Despite the assumed stress and burden thought to be associated with managing a child with an intellectual disability, the mean FES results for families with a child with WS were within normal limits. For example, overall the families reported high levels of cohesion and support and low levels of conflict. However, there is evidence of variability across most domains of the FES as seen in the range of scores, with some family’s ratings being either more than or less than one standard deviation below or above the standardised mean. For example, the results indicated that some families experience lower levels of control and organisation. Some aspects of the family environment were found to be related to higher outcomes in adaptive abilities. Overall, adaptive behaviour (ABC), Socialisation and Daily Living Skills were significantly higher for individuals whose families indicated that they participated in more social and recreational activities outside of school or work (for example, socialising with friends, taking part in sports, going to movies, having a hobby, attending a course or lesson of interest outside of school) and intellectual and cultural activities (for example attending plays or concerts, enjoying music, art, literature). It may be through taking part in these activities, that these families are providing more learning and enrichment opportunities for their child with WS which may, in turn, result in their being able to develop higher abilities. On the other hand, it may be that families with higher functioning individuals or more financial resources are better able to participate in such activities. However, no significant correlations were evident between the family's level of socioeconomic status (SES) or their level of intellectual functioning and FES ratings. Further research is needed to determine if there are any barriers which may prevent families from being able to take part in these activities (e.g., lack of facilities in their area).

While there was no relationship between intellectual/cultural and active/recreational activities and the maladaptive behaviour scales, it is possible that some behavioural issues may reduce the families' ability to take part in these sorts of activities. Higher levels of externalising problem behaviours were associated with lower scores on the Independence scale which measures the family's emphasis on self-sufficiency and making independent decisions. The most common externalising problems reported were for impulsivity and problems with inhibition. It may be the case that higher levels of these sorts of problem behaviours may make families less willing to encourage individuals to be self-sufficient. Alternatively, it may be that families are unaware of the potential benefits of such activities. These are areas which require further investigation. In addition, families that scored high on Control (which indicated that they used set rules, routines and procedures to run family life) were also associated with higher overall adaptive abilities and higher Daily Living Skills. The use of set routines and procedures may assist individuals with WS by providing the repetition and practice required to develop and master these skills. These findings suggest that independence in everyday living skills may be enhanced for individuals with WS by providing appropriate structure and routine, not only at home, as the home is the most common environment for adults with WS (Elison, Stinton & Howlin, 2010), but also within other environments. As noted previously, the school environment automatically provides this structure and the opportunity for social interactions. Therefore, outside of school and beyond formal school, individuals need to be challenged and stimulated with appropriate activities such as work and recreational pursuits.

Clinical Implications

Understanding functional impairments in WS is required in order to diagnose the level of intellectual disability and to provide informed opinions with regard to their daily vocational and vocational support needs. However, an important message from this research is that, due to the evidence of heterogeneity in the adaptive and cognitive skills of individuals with WS, it is inappropriate to rely solely on a global score. The current findings highlight that a mean score of adaptive functioning and a single FSIQ score are unlikely to accurately reflect the support needs for a WS individual. Rather, a comprehensive neuropsychological assessment is required to develop an adequate understanding of an individual's pattern of relative strengths and weaknesses across the domain areas of conceptual, social and practical skills in addition to cognitive strengths and weaknesses. Such a detailed understanding is also required to assist in educational and vocational planning (Harrison & Boney, 2002).

Having knowledge about the individual's adaptive and cognitive strengths and weaknesses can help to guide appropriate teaching and learning strategies, for example whether they would benefit from modelling, rehearsal, distributed practice, coaching using verbal instruction etc. As demonstrated, the family environment is also an important part of intervention planning and parental attitudes and expectations towards how much responsibility they expect their child to undertake would need to be evaluated and guidance provided as to reasonable expectations for adaptive skills depending on the child's age and cognitive strengths and weaknesses.

Weaknesses in adaptive functioning will need to be further analysed to determine whether the individual either does not have the skill or, alternatively, has the ability but does not use it when required (Harrison & Boney, 2002). Lack of ability may be due to several contributing factors, including a specific cognitive problem, such as a visuo-spatial or motor deficit or simply not having been taught that skill. Whereas, having the skill, but not using it when it is required may be due to behavioural, psychological or cognitive issues such as low frustration tolerance, anxiety or inattention. Therefore, it is important for families and education and health professionals to have a sound knowledge about how the WS individual's behavioural, psychological or cognitive difficulties may impact on their development of everyday skills.

As noted earlier, the revised Diagnostic and Statistical Manual of Mental Disorders fifth edition (DSM-5; APA, 2013) has placed a greater emphasis on level of adaptive functioning in the diagnosis of an intellectual disability with a move away from a single IQ score to determine the level of severity of intellectual disability (mild, moderate, severe, profound). Impairments in adaptive functioning and the subsequent level of support that the individual is likely to require for day-to-day activities is now the specifier for level of impairment. Also, due to the evidence that adaptive skill relative to same age peers plateaus with age, the severity level of the intellectual impairment for individuals with WS may change over time. This means that WS individuals will require regular reviews, especially as they move from childhood to adulthood, as their support needs are likely to change and, in particular, increase. As noted by Fu (2012), adaptive needs may be different at different life stages and this may certainly be the case in WS. It will be important for families to understand that this will not indicate a decline in intellectual abilities, as IQ is relatively stable over time, rather it reflects an increased need for support and assistance. Conversely, improvements in adaptive functioning following intervention which leads to the acquisition of skills which are shown to be stable and generalise to different situations can result in improvement in the classification of intellectual disability (APA, 2013).

Limitations and Future Directions

Findings from the current study must be considered within the context of a number of methodological limitations. Firstly, the small sample size. While the sample size in the current study is comparable to many other studies in the area, and while we acknowledge the practical limitations of studying such a rare disorder, larger sample sizes would be of great benefit to allow for the opportunity to explore a number of measurement variables without compromising the power of finding statistically significant differences.

Another limitation of the study is its cross-sectional design. Findings will be strengthened by longitudinal follow ups, including a longitudinal follow up of participants in the current study, to more fully understand changes in adaptive functioning and relative strengths and weaknesses over time. Of particular interest will be whether global adaptive functions and, more specifically, Communication difficulties and Daily Living Skills fail to make age appropriate gains over time in the current WS cohort.

In addition, future studies would benefit from additional informant ratings, such as teacher reports or reports by both parents independently to allow for different contexts and different viewpoints. It is important to obtain information from parents, caregivers, and teachers to help prioritise intervention goals to ensure that the highest priority is given to skills which are likely to have the most impact in everyday life (Harrison & Boney, 2002).

The current study also lacked a control group. While comparisons to other neurodevelopmental syndromes will be of import in future studies, the use of typically developing control groups, such as sibling controls, would also be beneficial. Some studies have compared WS to other neurodevelopmental syndromes. These studies do suggest syndrome-specific patterns, indicating that the patterns observed in WS are not simply reflecting intellectual disability more generally. The use of siblings would control for family environment characteristics, providing a unique opportunity to compare WS and typically developing individuals within the same home environments.

The major strength of the study was the genetic homogeneity of the group. Future research into phenotypes of WS will be enhanced by all researchers conducting full genotyping of their participants to ensure samples are homogenous. Comparisons between Porter at al., (2009) and the current study highlight this case in point (see above).

Concluding paragraph

This study examined adaptive functioning in a group of WS individuals with the standard ~1.6Mb genetic deletion. This was the first study to use the interview version of the Vineland Adaptive Behavior Scale – Second Edition (Vineland II, Sparrow et al., 2005), which provides adaptive functioning levels at the global, domain and subdomain levels. This study was also the first to investigate the relationship between family environment characteristics and an individual's adaptive functioning skills.These findings highlight the need to explore individual profiles of cognitive, adaptive and maladaptive impairment in WS and to explore the possibility of scatter at the subdomain and/or domain levels, which may render higher-level or summary scores uninterpretable. Chronological age and the family environment also need to be considered when exploring adaptive functions in WS. Although preliminary, these findings suggest the need for greater social integration and more structured and enriched environments for adults with WS in order to maintain their childhood levels of daily independence.

References

Achenbach, T.M., & Rescorla, L.A. (2001). *Manual for the ASEBA School-Age Forms & Profiles.* Burlington, VT: University of Vermont, Research Centre for Children, Youth & Families.

American Psychiatric Association. (2000). *Diagnostic and statistical manual of mental disorders* (4th ed., Text Rev.). Washington, DC: Author.

American Psychiatric Association. (2013). *Diagnostic and statistical manual of mental disorders* (5th ed.). Washington, DC: Author.

Australian Bureau of Statistics. (2006). *Census of Population and Housing: Index of Relative Socio-Economic Disadvantage (IRSD).* Retrieved from http://www.abs.gov.au/AUSSTATS/abs@.nsf/DetailsPage/2033.0.55.0012006?OpenDocument.

Bellugi, U., Lichtenberger, L., Jones, W., Lai, Z., & St. George, M. (2000). The neurocognitive profile of Williams syndrome: A complex pattern of strengths and weaknesses. *Journal of Cognitive Neuroscience, 12,* 7-29.

Bracken, B.A. (1988). Ten psychometric reasons why similar tests produce dissimilar results. *Journal of School Psychology, 26,* 155-166.

Bradley, R.H. (1993). Children's home environments, health, behaviour, and intervention efforts: A review using the HOME inventory as a marker measure. *Genetic, Social, and General Psychology Monographs, 119,* 437-490.

Brock, J. (2007). Language abilities in Williams syndrome: A critical review. *Development and Psychopathology, 19,* 97-127.

Bruininks, R.H., Woodcock. R.W., Weatherman, R.F., & Hill, B.K. (1996). *Scales of Independent Behavior – Revised.* Itasca, IL: Riverside Publishing Company.

Cherniske, E.M., Carpenter, T.O., Klaiman, C., Young, E., Bregman, J., Insogna, K., Schultz, R.T., & Pober, B.R. (2004). Multisystem study of 20 older adults with Williams syndrome. *American Journal of Medical Genetics, 131A,* 255-264.

Cohen, J. (1988). *Statistical power analysis for the behavioral sciences* (2nd ed.). Hillsdale, NJ: Lawrence Earlbaum Associates.

Davies, M., Howlin, P., & Udwin, O. (1997). Independence and adaptive behaviour in adults with Williams syndrome. *American Journal of Medical Genetics, 70,* 188-195.

De Bildt, A., Systema, S., Kraijer, D., Sparrow, S., & Minderaa, R. (2005). Adaptive functioning and behaviour problems in relation to level of education in children and adolescents with intellectual disability. *Journal of Intellectual Disability Research, 49,* 672-681.

Dimitropoulos, A., Ho, A.Y., Klaiman, C., Koenig, K., Schultz, R.T. (2009). A comparison of behavioural and emotional characteristics in children with autism, Prader-Willi syndrome, and Williams syndrome. *Journal of Mental Health Research in Intellectual Disabilities, 2,* 220-243.

Di Nuovo, S., & Buono, S. (2011). Behavioral phenotypes of genetic syndromes with intellectual disability: Comparison of adaptive profiles. *Psychiatry Research, 189,* 440-445.

Dixon, D.R. (2007). Adaptive behaviour scales. *International Review of Research in Mental Retardation, 34,* 99-140.

Dodd, H.F., & Porter, M.A. (2009). Psychopathology in Williams syndrome: The effect of individual differences across the life span. *Journal of Mental Health Research in Intellectual Disabilities, 2,* 89-109.

Donnai, D. & Karmiloff-Smith, A. (2000). Williams syndrome: From genotype through to the cognitive phenotype. *American Journal of Medical Genetics, 97,* 164-171.

Dykens, E.M. (2003). Anxiety, fears, and phobias in persons with Williams syndrome. *Developmental Neuropsychology, 23,* 291-316.

Dykens, E.M. & Hodapp, R.M. (2007). Neuropsychiatric genetic syndromes: Three steps toward improving the measurement of behavior in behavioral phenotype research. *Child and Adolescent Psychiatric Clinics of North America, 16,* 617-630.

Einfeld, S.L., & Tonge, B.J. (2002). *Manual for the Developmental Behaviour Checklist* (2nd ed.) – *Primary Carer Version (DBC-P) and Teacher Version (DBC-T).* Sydney, Australia: Monash University Centre for Developmental Psychiatry and Psychology and School of Psychiatry, University of New South Wales, Australia.

Einfeld, S.L., Tonge, B.J., & Florio, T. (1997). Behavioral and emotional disturbance in individuals with Williams syndrome. *American Journal on Mental Retardation, 102,* 45-53.

Elison, S., Stinton, C., & Howlin, P. (2010). Health and social outcomes in adults with Williams syndrome: Findings from cross-sectional and longitudinal cohorts. *Research in Developmental Disabilities, 31,* 587-599.

Ewart, A.K., Morris, C.A., Atkinson, D., Jin, W., Sternes, K., Spallone P., Stock, A.D., Leppert, M., & Keating, M.T. (1993). Hemizygosity at the elastin locus in a developmental disorder, Williams syndrome. *Nature Genetics, 5,* 11–16.

Fisch, G.S., Carpenter, N.J., Holden, J.J.A., Simensen, R., Howard-Peebles, P.N., Maddalena, A., Pandya, A., & Nance, W. (1999). Longitudinal assessment of adaptive and maladaptive behaviors in Fragile X males: Growth, Development, and Profiles. *American Journal of Medical Genetics, 83,* 257-263.

Fisch, G.S., Carpenter, N., Howard-Peebles, P.N., Holden, J.J.A., Tarleton, J., Simensen, R., Nance, W. (2007). Research review: Studies of age-correlated features of cognitive-behavioral development in children and adolescents with genetic disorders. *American Journal of Medical Genetics Part A, 143A,* 2478-2489.

Fisch, G.S., Carpenter, N., Howard-Peebles, P.N., Holden, J.J.A., Tarleton, J., & Simensent, R. (2010). The course of cognitive-behavioral development in children with FMR1 Mutation, Williams-Beuren syndrome, and Neurofibromatosis Type 1: The effect of gender. *American Journal of Medical Genetics Part A, 152A,* 1498-1509.

Fisch, G.S. (2000). Psychology genetics. *American Journal of Medical Genetics (Seminar Medical Genetics), 97,* 109-111.

Fu, J.T. (2012). *The association of intelligence, visual-motor functioning, and personality characteristics with adaptive behavior in individuals with Williams syndrome.* (Order No. 3524353, Alliant International University). *ProQuest Dissertations and Theses,* 117. Retrieved from http://search.proquest.com/docview/1080530412?accountid=12219. (1080530412).

Glaser, B., Hessl, D., Dyer-Friedman, J., Johnston, C., Wisbeck, J., Taylor, A., & Reiss, A. (2003). Biological and environmental contributions to adaptive behaviour in Fragile X syndrome. *American Journal of Medical Genetics, 117A,* 21-29.

Gosch, A., & Pankau, R. (1994). Social-emotional and behavioural adjustment in children with Williams syndrome. *American Journal of Medical Genetics, 53,* 335-339.

Greer, M.K., Brown III, F.R., Pai, G.S., Choudry, S.H., & Klein, A.J. (1997). Cognitive, adaptive, and behavioral characteristics of Williams syndrome. *American Journal of Medical Genetics (Neuropsychiatric Genetics), 74,* 521-525.

Harrison, P.L. (1987). Research with adaptive behaviour scales. *The Journal of Special Education, 21,* 37-68.

Harrison, P.L., & Boney, T.L. (2002). Best practices in the assessment of adaptive behavior. In A. Thomas & J. Grimes (Eds), *Best Practices in School Psychology-IV* (pp. 1167-1179). Washington, DC: National Association of School Psychologists.

Harvill, L.M. (1991). Standard Error of Measurement. *Educational Measurement: Issues and Practice, 10,* 33-41.

Hauser-Cram, P., Warfield, M.E., Shonkoff, J.P., Krauss, M.W., Upshur, C.C., & Sayer, A. (1999). Family influences on adaptive development in young children with Down syndrome. *Child Development, 70,* 979-989.

Howell, A., Hauser-Cram, P., & Kersh, J.E. (2007). Setting the stage: Early child and family characteristics as predictors of later loneliness in children with developmental disabilities. *American Journal on Mental Retardation, 112,* 18-30.

Howlin, P., Davies, M., & Udwin, O. (1998). Cognitive functioning in adults with Williams syndrome. *Journal of Child Psychology & Psychiatry, 39,* 183-189.

Howlin, P., Elison, S., Udwin, O., & Stinton, C. (2010). Cognitive, linguistic and adaptive functioning in Williams syndrome: Trajectories from early to middle adulthood. *Journal of Applied Research in Intellectual Disabilities, 23,* 322-336.

Jarrold, C., Baddeley, A.D., & Hewes, A.K. (1998). Verbal and non-verbal abilities in the Williams syndrome phenotype: Evidence for diverging developmental trajectories. *Journal of Child Psychology and Psychiatry, 39,* 511-523.

John, A.E., & Mervis, C.B. (2010). Sensory modulation impairments in children with Williams syndrome. *American Journal of Medical Genetics Part C (Seminars in Medical Genetics), 154C,* 266-276.

Jones, W., Bellugi, U., Lai, Z., Chiles, M., Reilly, J., Lincoln, A., & Adolphs, R. (2000). Hypersociability in Williams Syndrome. *Journal of Cognitive Neuroscience, 12:Supp,* 30-46.

Keith, T.Z., Fehrmann, P.G., Harrison, P.L., & Pottebaum, S.M. (1987). The relation between adaptive behaviour and intelligence: Testing alternative explanations. *Journal of School Psychology, 25,* 31-45.

Kennedy, J.C., Kaye, D.L., & Sadler, L.S. (2006). Psychiatric diagnoses in patients with Williams syndrome and their families. *Jefferson Journal of Psychiatry, 20,* 22-31.

Laws, G., & Bishop, D.V.M. (2004). Pragmatic language impairment and social deficits in Williams syndrome: A comparison with Down's syndrome and specific language impairment. *International Journal of Language & Communication Disorders, 39,* 45-64.

Leyfer, O. T., Woodruff-Borden, J., Klein-Tasman, B. P., Fricke, J. S., & Mervis, C.B. (2006). Prevalence of psychiatric disorders in 4 to 16-year-olds withWilliams syndrome. *American Journal of Medical Genetics, Part B:(Neuropsychiatric Genetics), 141B*, 615-622.

Liss, M., Harel, B., Fein, D., Allen, D., Dunn, M., Feinstein, C., Morris, R., Waterhouse, L., & Rapin, I. (2001). Predictors and correlates of adaptive functioning in children with developmental disorders. *Journal of Autism and Developmental Disorders, 31,* 219-230.

Martens, M.A., Wilson, S.J., & Reutens, D.C. (2008). Research review: Williams syndrome: A critical review of the cognitive, behavioral, and neuroanatomical phenotype. *The Journal of Child Psychology and Psychiatry, 49,* 576-608.

Mather, N. & Woodcock, R.W. (2001). Examiner's Manual. *Woodcock-Johnson III Tests of Cognitive Abilities.* Rolling Meadows, IL: Riverside Publishing.

McGrew, K., & Woodcock, R. W. (2001). Technical Manual. *Woodcock-Johnson III.* Rolling Meadows, IL: Riverside Publishing.

Mervis, C.B., & John, A.E. (2010). Cognitive and behavioral characteristics of children with Williams syndrome. *American Journal of Medical Genetics Part C (Seminars in Medical Genetics), 154C*, 229-248.

Mervis, C.B., & Klein-Tasman, B.P. (2000). Williams syndrome: Cognition, personality, and adaptive behavior. *Mental Retardation and Developmental Disabilities Research Reviews, 6,* 148-158.

Mervis, C.B., Klein-Tasman, B.P., & Mastin, M.E. (2001). Adaptive behavior of 4- through 8-year old children with Williams syndrome. *American Journal on Mental Retardation, 106,* 82-93.

Meyer-Lindenberg, A., Mervis, C.B., & Berman, K.F. (2006). Neural mechanisms in Williams syndrome: A unique window to genetic influences on cognition and behaviour. *Nature Reviews Neuroscience, 7,* 380-393.

Mink, I.T., Nihira, K., & Meyers, C.E. (1983). Taxonomy of family life styles: Homes with TMR children. *American Journal of Mental Deficiency, 87,* 484-497.

Moos, R.H., & Moos, B.S. (1994). *Family Environment Scale Manual* (Third Edition). Palo Alto, CA: Consulting Psychologists Press.

Morris, C.A. (2010). Introduction: Williams syndrome. *American Journal of Medical Genetics Part C (Seminars in Medical Genetics), 154C,* 203-208.

Pezzini, G., Vicari, S., Voltera, V., Milani, L., & Ossella, M.T. (1999). Children with Williams syndrome: Is there a single neuropsychological profile? *Neuropsychologia, 15,* 141-155.

Phillips, K.D. (2008). *Emotion regulation and dysregulation in children and adolescents with Williams syndrome* (Doctoral dissertation). Retrieved from ProQuest database. (304453044).

Plomin, R. & Thompson, L.A. (1993). Genetics and high cognitive ability. *CIBA Foundation Symposium, 178,* 67-79.

Porter, M.A. & Coltheart, M., (2005). Cognitive heterogeneity in Williams syndrome. *Developmental Neuropsychology, 27,* 275-306.

Porter, M.A., Dobson-Stone, C., Kwok, J.B.J., Schofield, P.R., Beckett, W., & Tassabehji, M. (2012). A role for transcription factor GTF2IRD2 in executive function in Williams-Beuren syndrome. *PLoS ONE, 7,* e47457. DOI: 10.1371/journal.pone.0047457.

Porter, M. & Dodd, H. (2011). A longitudinal study of cognitive abilities in Williams syndrome. *Developmental Neuropsychology, 36,* 255-272.

Porter, M.A., Dodd, H., & Cairns, D. (2009). Psychopathological and behaviour impairments in Williams-Beuren syndrome: The influence of gender, chronological age, and cognition. *Child Neuropsychology, 15,* 359-374.

Riby, D.M., & Porter, M.A. (2010). Chapter 5 – Williams syndrome. *Advances in Child Development and Behavior, 39,* 163-209.

Rothman, K.J. (1990). No adjustments are needed for multiple comparisons. *Epidemiology, 1*, 43-46.

Rowe, M.L. (2007). *Mastery motivation in young children with Williams syndrome or Down syndrome* (Order No. 3267110, University of Louisville). *ProQuest Dissertations and Theses*, 131. Retrieved from http://search.proquest.com/docview/304837706?accountid=12219. (304837706).

Sattler, J.M. (1992). *Assessment of children: Revised and Updated* (3rd ed.). San Diego, CA: Jerome M. Sattler, Publisher. Inc.

Sattler, J.M. (2001). *Assessment of children: Cognitive applications* (4th ed.). San Diego, CA: Jerome M. Sattler, Publisher. Inc.

Schrank, F.A., Flanagan, D.P., Woodcock, R.W., & Mascolo, J.T. (2002). *Essentials of WJ III Cognitive Abilities Assessment.* New York: John Wiley & Sons.

Schrank, F. A., & Woodcock, R. W. (2009). WJ III Normative Update Compuscore and Profiles Program, *Australian Adaptation;* [WJ III NU (AA)], [Computer software]. Adaptation of the *Woodcock-Johnson III Normative Update* (WJ III NU) Compuscore and Profiles Program. Rolling Meadows, IL: Riverside Publishing.

Searcy, Y.M., Lincoln, A.J., Rose, F.E., Klima, E.S., Bavar, N., & Korenberg, J.R. (2004). The relationship between age and IQ in adults with Williams syndrome. *American Journal on Mental Retardation, 109,* 231-236.

Sparrow, S., Balla, D., & Cicchetti, D. (1984). *Vineland Adaptive Behavior Scales. Survey Forms Manual.* Circle Pines, MN: American Guidance Services.

Sparrow, S.S., Cicchetti, D.V., & Balla, D.A. (2005). *Vineland Adaptive Behavior Scales* (2nd Ed.). Survey Forms Manual. AGS Publishing, Circle Pines, MN: AGS Publishing.

Strømme, P., Bjørnstad, P.G., Ramstad, K. (2002). Prevalence estimation of Williams syndrome. *Journal of Child Neurology, 17,* 269-271.

Tassabehji, M., Metcalfe, K., Karmiloff-Smith, A., Carette, M.J., Grant, J., Dennis, N., et al. (1999). Williams syndrome: Use of chromosomal microdeletions as a tool to dissect cognitive and physical phenotypes. *American Journal of Human Genetics, 64,* 118-125.

Tasse, M.J., Schalock, R.L., Balboni, G., Bersani, H., Borthwick-Duffy, S.A., Spreat, S., Thissen, D., Widaman, K.F., & Zhang, D. (2012). The construct of adaptive behavior: Its conceptualization, measurement, and use in the field of intellectual disability. *American Journal on Intellectual and Developmental Disabilites, 117*, 291-303.

Udwin, O. & Yule, W. (1991). A cognitive and behavioural phenotype in Williams syndrome. *Journal of Clinical and Experimental Neuropsychology, 13*, 232-244.

Vineland II ASSIST Scoring and Reporting System for Survey Forms [Computer software]. (2008). San Antonio, TX: Pearson.

Warfield, M.E. (1995). *Early predictors of child behaviour problems: Examining the family environment.* Paper presented at the biennial meeting of the Society for Research in Child Development, Indianapolis, IN.

Widaman, (2010). Review of the Vineland Adaptive Behavior Scales, Second Edition. In R.A. Spies, J.F. Carlson, & K.F. Geisinger (Eds.), *The eighteenth mental measurements yearbook* (pp. 682-684). Lincoln, NE: University of Nebraska Press.

Woodruff-Borden, J., Kistler, D.J., Henderson, D.R., Crawford, N.A., & Mervis, C.B. (2010). Longitudinal course of anxiety in children and adolescents with Williams syndrome. *American Journal of Medical Genetics Part C: Seminar in Medical Genetics, 154C*, 277-290.

Woodcock, R.W., McGrew, K.S., & Mather, N. (2001). *Woodcock-Johnson III Tests of Cognitive Abilities.* Rolling Meadows, IL: Riverside Publishing.

Appendix

WJ III COG Standard Battery (Tests 1-10)

1. Verbal Comprehension: Comprised of four subtests. Picture Vocabulary involves naming pictured objects; Synonyms requires hearing a word and saying a word similar in meaning; Antonyms requires hearing a word and saying a word opposite in meaning; Verbal Analogies measures ability to reason using lexical knowledge.

2. Visual-Auditory Learning: Long-term storage and retrieval. Measures the ability to learn, store and retrieve a series of visual-auditory associations. The subject is asked to associate new visual symbols (rebuses) with familiar words and to then translate a series of symbols presented as a reading passage.

3. Spatial Relations: Visual-spatial thinking. The subject has to identify the two or three pieces that combine to make a target shape.

4. Sound Blending: Auditory processing. After hearing parts of a word (syllables or phonemes) the subject has to blend the sounds into a word.

5. Concept Formation: Fluid reasoning. Measures ability to identify and state the rule for a concept about a set of coloured geometric figures.

6. Visual Matching: Processing speed. Individual has to locate and circle two identical numbers in a row of six numbers and has a 3-minute time limit.

7. Numbers Reversed: Short term memory. The individual has to hold a span of numbers in immediate awareness while performing mental operation of reversing the sequence.

8. Incomplete Words: Auditory processing. The individual hears a word with one or more phonemes missing and they have to identify the complete word.

9. Auditory Working Memory: Short-term auditory memory span, working memory and divided attention. Individual is presented with a series of words and digits and has to rearrange the information by repeating the objects in sequential order and then the numbers in sequential order.

10. Visual-Auditory Learning Delayed: Long-term retrieval. Measures the subject's ability to recall the symbols (rebuses) present in Task 2 (Visual-Auditory Learning).

WJ III COG Extended Battery (Tests 11-20)

11. General Information: Measures general knowledge of common characteristics of certain objects. There are two components: (1) the 'where' component, "where would you find…(an object)?" and (2) the 'what' component where individuals are asked "what would you do with…(an object)?"

12. Retrieval Fluency: Long-term retrieval. Fluency of retrieval from stored knowledge. Subject is asked to name as many examples from a given category within a 1-minute time period.

13. Picture Recognition: Visual memory of objects or pictures. Subject is asked to recognise a subset of previously presented pictures from a set of distracting pictures.

14. Auditory Attention: Speech sound discrimination requiring selective attention. Subject listens to a word while seeing four pictures and has to point to the correct picture for the word. Measures ability to discriminate similar sounding words from an audio recording with an increasing level of background noise as a distracter.

15. Analysis-Synthesis: Fluid reasoning. Measures the ability to analyse the components of an incomplete logic puzzle and use reasoning to draw conclusions about the missing components.

16. Decision Speed: Processing speed. Ability to locate quickly the two pictures that are most similar conceptually within a 3-minute time limit.

17. Memory for Words: Short-term auditory memory span. Subject is asked to repeat lists of unrelated words in the correct sequence.

18. Rapid Picture Naming: Cognitive fluency. In a 2-minute time limit, rapidly identify and name pictures of common objects.

19. Planning: Executive processing. Requires individuals to plan a tracing route over a dotted line drawing adhering to the rules that they cannot lift their pencil nor trace over the same segment twice. Requires 'forward thinking' to plan a sequence of steps prior to initialising action.

20. Pair Cancellation: Executive processing, attention/concentration, processing speed. Individual is presented with rows which contain repeating pictures of a dog and a ball in random order. Subject is asked to locate and mark a repeated pattern as quickly as possible in a 3-minute time period.

Note. GIA-Standard (GIA-Std) scale consists of seven tests (Tests 1 to 7).

GIA-Extended (GIA-Ext) scale consists of fourteen tests (Tests 1-7 plus Tests 11-17).

Brief Measure of Intellectual Ability (BIA) consists of Test 1, Test 5 and Test 6

Functional Basic Reading Skills in Williams Syndrome

Gabrielle Brawn, Saskia Kohnen, Melanie Porter

Macquarie University

Correspondence should be sent to Melanie A. Porter, Psychology Department, Macquarie University, Sydney, Australia 2109. Phone: +61 2 9850 6768, Fax: +61 2 9850 8062, E-mail: melanie.porter@mq.edu.au

Abstract

In this study we investigated whether individuals with William syndrome (WS) can attain a functional level of basic reading skills. The broad aims were to 1) explore heterogeneity in reading by using a cognitive neuropsychological approach based on Dual Route theory to profile each individual's reading performance, 2) investigate whether a functional level of reading ability is associated with adaptive functioning or everyday living skills in WS, and 3) consider the relationship between reading, overall intellectual functioning (IQ) and cognitive skills in WS. Thirty individuals with WS (mean chronological age 21 years and mean mental age 7 years 7 months) were asked to read regular words, irregular words and nonwords from the Castles and Coltheart Reading Test 2 (CC2). Each individual's performance on irregular word and nonword reading was analysed to determine relative strengths and weaknesses in reading via the lexical (whole-word) versus nonlexial (decoding) pathway. Overall intellectual ability and cognitive skills were assessed on the Woodcock Johnson 3rd Edition Tests of Cognitive Abilities (WJ III COG). Parent or caregiver completed an interview of adaptive functioning using the Vineland Adaptive Behavior Scales (Vineland II). The results confirmed the heterogeneity of the reading profile in WS and demonstrated that higher reading ability was found to be associated with increased outcomes in adaptive functioning, in particular Written and Expressive communication skills and Community Living skills highlighting the potential benefits of developing reading abilities in WS. While IQ was not found to be a determining factor in reading performance, several cognitive skills known to be related to reading ability in typically developing individuals were found to be predictive of reading performance. Implications for appropriate reading instruction are discussed.

Introduction

Literacy is an important skill which enables an individual to participate more independently within society. Developing literacy is no less important for individuals with an intellectual disability as a means to contribute to their independence and quality of life (Laing, 2002). The concept of ‘functional literacy’ refers to a level of reading ability which would allow a person to cope with the literacy demands of many everyday tasks, such as reading a bus timetable, television guide or instructions on a medicine bottle. Literacy skills also provide opportunities for leisure activities (for example, reading a book, magazine, newspaper) and allow individuals to make use of modern day technology for social and/or communication purposes such as email, text messages or social media. Investigating literacy abilities in individuals with Williams syndrome (WS), who typically have a mild to moderate intellectual disability is, therefore, important, but has received less attention within the WS literature than other cognitive and intellectual abilities. While ultimately reading is about gaining information from written text (“reading comprehension”), this study will focus on single-word reading ability, as this can be considered a foundation skill upon which other abilities, such as reading comprehension, depend. In addition, we explored the relationship between functional level reading ability and adaptive functioning to investigate the impact that developing literacy skills can have on everyday functioning.

Williams syndrome (WS), also known as Williams-Beuren syndrome is a relatively rare genetic disorder caused by a microdeletion of approximately 26 genes on chromosome 7q11.23, including the elastin gene (Ewart et al., 1993; Lowery et al., 1995). There are reports of smaller and larger gene deletions in WS (e.g., see Heller, Rauch, Lüttgen, Schröder, & Winterpacht, 2003; Karmiloff-Smith et al., 2012; Tassabehji, 2003). Prevalence has been estimated to be around 1 in 7,500 (Strømme, Bjørnstad, & Ramstad, 2002). Mean level of intellectual functioning (IQ) is typically from 50 to 60 (see review by Martens, Wilson, & Reutens, 2008) with some studies reporting a higher verbal intellect to nonverbal intellect (e.g., Howlin, Davies, & Udwin, 1998; Udwin & Yule, 1991; see Martens et al., 2008). However, this discrepancy may be attributable to a significant weakness in visuo-spatial abilities (Brock, 2007). The cognitive profile is also associated with relative strengths in picture vocabulary and verbal short-term memory (see review by Mervis & John, 2010). There is also evidence of heterogeneity in intellectual functioning and cognitive abilities

(Jarrold, Baddeley, & Hewes, 1998; Pezzini, Vicari, Voltera, Milani & Ossella, 1999; Porter & Coltheart, 2005, Tassabehji et al., 1999).

Conners, Moore, Loveall, and Merrill (2011) reviewed the memory profiles of individuals with WS and the implication for reading development. It was noted that individuals with WS have weaknesses in: spatial memory, visual-auditory learning (associative learning) and semantic retrieval, while relative strengths included: immediate verbal and visual recall, visual delayed memory and learning and phonological retrieval. Relative strengths in visual memory were thought to be likely to help children with WS develop a sight vocabulary. Visual memory has been found to be significantly and positively correlated with sight word learning in typically developing five year olds, particularly when letter-sound knowledge was poor (Stuart, Masterson, & Dixon, 2000). Relative strengths in verbal working memory and phonological retrieval could offset weakness in associative learning. Conners et al. (2011) stated that due to within-syndrome variability, instructional approaches should be tailored to the individual by providing extra support in weak areas and by finding ways to capitalise on strengths (e.g., see Riby & Porter, 2010).

As genetic and cognitive variability has been demonstrated in WS, it is also important to consider heterogeneity in reading ability. If different reading difficulties can be identified, this will be of clinical value to enable identification of appropriate teaching methods for the individual rather than syndrome specific recommendations.

Reading Ability in Williams Syndrome

Studies that have assessed reading ability in WS have included individuals across a wide age range, with the youngest being around 9 years of age through to the oldest adults aged around 39 years. These studies have consistently found that reading ability in WS is typically low, with the mean reading age equivalency ranging from 6 years 5 months for a group with mean chronological age (CA) of 15 years 1 month (Laing, Hulme, Grant, & Karmiloff-Smith, 2001) through to 8 years 8 months for a group with mean CA of 21 years 9 months (Udwin, Davies, & Howlin, 1996). Consistent with findings of cognitive heterogeneity in WS, there is also evidence of a wide range of abilities in reading skills. For example, Udwin et al.'s (1996) study of 62 WS adults aged from 19 to 39 years reported that reading ages ranged from 6 through to 18 years (also see Becerra, John, Peregrine & Mervis, 2008 as cited in Mervis, 2009).

Several studies also included some individuals that could not read any words at all (e.g., Howlin et al., 1998; Laing et al., 2001; Levy & Antebi, 2004; Levy, Smith, & Tager-Flusberg, 2003; Udwin, Yule, & Martin, 1987). In summary, average reading levels of individuals with WS tend to be significantly lower than chronological age and approximately equivalent to a 6 to 8 year age level. There is also evidence of considerable variability, with some individuals unable to read at all, while others read at a significantly higher level. What remains relatively unexplored to date is if this heterogeneity is related to other aspects of the participants' cognitive and intellectual profiles.

Case Study and Case Series Approaches

Given this variability, detailed single case studies and case series form an important aspect of the evidence base in reading in WS. Most published case studies with WS participants are based on Dual Route theory (e.g., Coltheart, Rastle, Perry, Langdon, & Ziegler, 2001). One of the main concepts of the Dual Route theory is that there are two ways to read: one relies on applying the letter-to-sound rules (called nonlexical reading), while the other relies on accessing a stored mental representation of the word (called lexical reading). Words which follow the letter-to-sound rules (pronounceable nonwords and regular words) can be read by the nonlexical pathway, while irregular words can only be read via the lexical pathway (Castles et al., 2009; Coltheart et al., 2001).

Two studies (Barca, Bello, Volterra, & Burani, 2009; Menghini, Verucci, & Vicari, 2004) have indicated relative weaknesses in the nonlexical reading route in comparison to the lexical route. Temple (2003) reported the case of a child with deep dyslexia; this child was unable to read nonwords and made semantic errors when reading words. In contrast to these cases with nonlexical reading difficulties, Temple (2006) reported that eight children with WS had nonword reading (via the nonlexical route) comparable to mental age matched controls, while their reading of irregular words (lexical route) was impaired. Another single case study (Dessalegn, Landau, & Rapp, 2012) identified visual-spatial difficulties in WS as being associated with visual orientation (e.g., pump → bump) and ordering (e.g., bowl → blow) errors. In summary then, using Dual Route theory to investigate reading ability, cases have indicated that individuals with WS seem to have a particular weakness in nonlexical compared to lexical reading, although there have also been reports of the reverse pattern.

Nonword Reading in WS

Given that the majority of case reports found relative weaknesses in nonlexical reading, we will provide a brief summary of group studies investigating nonword reading, which isolates the ability of the non-lexical route. Some studies have reported nonword reading in WS to be comparable to groups matched for reading age and receptive vocabulary (Laing et al, 2001) and mental age (Heinze & Vega, 2008; Temple, 2006), while other studies have found the opposite, with nonword reading significantly lower than controls matched on mental age (Menghini et al., 2004). There is obviously a wide range of abilities in nonword reading with some individuals unable to read any nonwords at all (Becerra et al., 2008 as cited in Mervis, 2009; Laing et al, 2001; Levy & Antebi, 2004; Levy et al., 2003), while others are able to read nonwords at an average or age appropriate level (Becerra et al., 2008 as cited in Mervis, 2009; Levy et al., 2003). This indicates that the nonlexical route or letter-sound rule-based route could be considered 'intact' for some WS individuals, while for others, it would be considered impaired.

Phonological and Phonemic Awareness

Phonological awareness is an oral language skill which includes segmenting and blending sounds in words. Phonological awareness has consistently been identified as being related to reading ability in typically developing children (Castles & Coltheart, 2004; Wagner & Torgesen, 1987; Windfuhr & Snowling, 2001). However, Castles and Coltheart's (2004) review did not find definitive evidence that PA skills precede the acquisition of reading ability. Similarly, specific training in phonemic awareness in pre-literate children was not found to lead to better acquisition of reading skills compared to those that did not receive PA training (Castles & Coltheart, 2004). Research with individuals with an intellectual disability (ID) has identified phonological processing skills to be associated with word recognition and nonword decoding abilities (Conners, Atwell, Rosenquist, & Sligh, 2001; Gombert, 2002; Verucci, Menghini, & Vicari, 2006; Wise, Sevcik, Romski, & Morris, 2010). However, the type of PA tasks utilised is an important consideration, as some tasks may place too great a demand on the meta-cognitive abilities for individuals with an intellectual disability (e.g., see Cupples &

Iacano, 2000). Studies have demonstrated that phonologically based reading instruction can be effective for children with Down syndrome (Cupples & Iacono, 2002) and children with intellectual disability more generally (Conners, Rosenquist, Sligh, Atwell, & Kiser, 2006). These findings suggest that children with intellectual disability can learn to read in ways similar to typically developing children and would, therefore, benefit from incorporating a phonics-based instruction when learning to read (Wise et al., 2010).

Several studies have found strong correlations between phonological or phonemic awareness tasks and reading in WS (e.g., Laing et al., 2001; Levy & Antebi, 2004; Levy et al., 2003; Menghini et al., 2004). These correlations made some authors conclude that an emphasis on the phonics approach to teaching reading should be appropriate for individuals with WS (e.g., Levy & Antebi, 2004; Levy et al., 2003). In contrast, Menghini et al. (2004) suggested that the WS participants had an impairment in grapheme-phoneme conversions and, therefore, recommended that reading instruction for WS should focus on whole word recognition given their ‘advantage’ in this area. They also recommend strengthening phonological awareness to improve grapheme-phoneme knowledge (but see Castles & Coltheart, 2004). Laing (2002) questioned whether programmes to teach reading in clinical populations should be syndrome specific or whether similar teaching strategies used with typically developing individuals would be suitable. In summary then, there is considerable disagreement in regards to a unitary recommendation for teaching procedures.

Reading and Adaptive Functioning

Overall then, reading abilities vary considerable between individuals with WS. However, for those individuals who can read it is important to consider how their reading abilities relate to their everyday functioning because this may help to determine the focus for educational instruction. If literacy skills are shown to help individuals cope with tasks associated with living in the community, then appropriate reading instruction to achieve this level would be essential (Bochner, Outhred, & Pieterse, 2001). We are only aware of one study that has included standardised measures of both reading ability and adaptive or everyday functioning. Howlin et al. (1998) assessed adaptive functioning on the Vineland Adaptive Behavior Scales (VABS; Sparrow, Balla, & Cicchetti, 1984) for 62 adults aged from 19 to 39 years. Their level of functioning across all adaptive domains was found to be low, with skills clustering around the 6 year old level. The mean word reading level (for 47 participants who could read) was 8 years 7 months (range 6 years through to 18 years). The authors noted that the individuals did not seem to be able to utilise their academic skills for their everyday functioning. Within the Communication domain, the Written subdomain was reported to be relatively higher than the Receptive or Expressive subdomains and the authors noted that this was surprising given their low level of spelling ability identified on testing. It should be noted that the VABS does not provide standard scores for the subdomains, only age equivalents, and comparison of age equivalents scores have statistical limitations due to the fact that the scale units are unequal (see Sparrow, Cicchetti, & Balla, 2005), making comparisons of these scores difficult. Thus, the current study aims to investigate the relationship between adaptive behaviour and literacy skills by using the updated Vineland Adaptive Behaviour Scales – 2nd Edition (Vineland II; Sparrow et al., 2005), which provides standardised subdomain scores which will allow investigation into how reading ability in WS impacts on everyday functioning.

Reading and Intellectual Functioning

In contrast to adaptive functioning, the relationship between overall intellectual functioning (Full Scale IQ or FSIQ) and verbal or perceptual intelligence with reading ability in WS has been investigated in several studies. An early study by Pagan, Bennett, LaVeck, Stewart, and Johnson. (1987) found that for all nine of their participants, reading scores were higher than FSIQ scores. The authors concluded that reading skills should be taught regardless of IQ level. However, other studies have indicated that reading ability is significantly related to IQ level in WS. In two related studies involving some of the same participants, Udwin et al. (1987; see also, Udwin et al., 1996) found that those able to attain a reading score were significantly older and had higher FSIQ’s than non-readers. Similarly, Howlin et al. (1998) reported that the non-readers had significantly lower FSIQ, verbal IQ (VIQ) and perceptual IQ (PIQ) than the group who could read. Levy et al. (2003) found that individuals with a FSIQ between 50 to70 were reading at their IQ level, whereas those with an IQ higher than 70 (only three participants) were reading within the low-average to average range (in many instances higher than their IQ level). For Hebrew-speaking adolescents, Levy and Antebi (2004) reported that their word reading was significantly correlated with IQ. The authors hypothesised that an IQ level within the impaired range was predictive of reading acquisition for individuals with WS, whereas in the general population, reading is not predicted on the basis of IQ (Levy et al., 2003).

In summary, there appears to be some evidence that reading ability is associated with intellectual ability for individuals with WS, which could be interpreted to mean that very low IQ explains poor reading. However, as has been shown with typically developing children, there is a need to look beyond overall intelligence to investigate specific reading processes to better understand the variation in reading abilities for those with intellectual disability (Conners et al., 2001).

Other Possible Correlates of Reading Ability

In addition to phonological awareness (PA) and general intellectual ability, other cognitive skills have been identified as being related to reading ability in typically developing children, including working memory, rapid automatic naming, receptive vocabulary and paired associative learning. Working memory (WM) is thought to be necessary for phonological decoding, as sounding phonological decoding requires the ability to hold each sound in mind temporarily while decoding the phonemes of the word and remember them long enough to then be able to blend the sounds together (Wagner & Torgesen, 1987; Strattman & Hodson, 2005). Both WM and PA have been shown to contribute uniquely to nonword reading (Strattman & Hodson, 2005). Rapid automatic naming (RAN) of familiar visual symbols (e.g., letters, digits, colours or objects) has been found to be correlated with reading ability; however the cause of the association remains under debate (Arnell, Klein, Joanisse, Busseri, & Tannock, 2009). Rapid naming may measure speed of lexical access (e.g. Wagner & Torgesen, 1987) and the strength of association between visual and phonological forms (Conners et al., 2011). Paired associative learning (Nilsen & Bourassa, 2008; Windfuhr & Snowling, 2001) is required for learning letter-sound associations and recognising words by their orthographic features.

Some studies reported an association between reading and RAN in WS (Laing et al., 2001), while other studies have not found correlations between reading and RAN (Dessalegn et al., 2012; Levy & Antebi, 2004; Levy et al., 2003). Levy et al. (2003) suggested that poor performance on RAN tasks may be related to specific difficulties with attention, slow verbal retrieval and visual-processing delays for individuals with

WS. Some studies have reported nonverbal abilities to be associated with reading ability in WS (Dessalegn et al., 2012; Laing et al., 2001; Levy et al., 2003). One study included an associative learning task (Laing et al., 2001) and found that WS participants were not influenced by the imageability of the word. Laing et al. suggested that this implies that semantic information plays a weaker role in reading in WS, however, Mervis (2009) counters that other studies, using typically developing children, have failed to find this semantic effect.

Overall, there is conflicting evidence in WS about the association between cognitive abilities known to be associated with reading in typically developing individuals, and whether the same abilities are related to reading ability in WS. Investigating these associations in WS is important not only theoretically, but also practically. From a theoretical point of view, it is interesting to know if populations with atypical cognitive profiles follow a different developmental trajectory from populations with typical cognitive profiles. From a practical point of view, these associations may have implications for the types of interventions that can be successful.

Aims of the Current Study

The current study had four aims. First, to investigate the reading profile of WS groups using the Castles and Coltheart Reading Test 2 (CC2; Castles et al., 2009), which looks at regular, irregular and nonword reading abilities independently (or lexical and nonlexical reading), to see if the majority of individuals with WS were functional readers, with a reading level equivalent to end of Year 5 primary level. Second, to determine whether reading ability was related to daily functional skills (measured using Vineland Adaptive Behaviour Scales – 2nd Edition, Vineland II; Sparrow et al., 2005) and whether reading abilities are on par with or greater than, or less than, FSIQ. Third, to investigate whether lexical and/or nonlexical reading ability was related to FSIQ or other cognitive abilities measured using the Woodcock-Johnson III Tests of Cognitive Abilities - Australian Adaptation (WJ III COG; Woodcock, McGrew, & Mather, 2001). Finally, to examine whether all individuals with WS showed a similar reading profile on the CC2 or whether there were distinct reading subtypes in WS, and if so, how this related to more specific cognitive abilities. The overarching aim was to see what percentage of individuals with WS were functional readers, with a reading level equivalent to end of Year 5 primary level and to provide information on possible avenues to assist non-functional WS readers.

Hypotheses

In relation to the first aim, based on previous research, we expected to find variability in the group's abilities, with some individuals able to read at a functional level while other individuals may not be able to read at all. Based on previous research, we expect to find the greatest difficulties with nonword/nonlexical reading. In terms of the second aim, although this has not been looked at before, we expect that the Written subdomain from the Vineland II would relate to reading ability on the CC2, in particular lexical reading, as the Written subdomain is a measure of functional reading and writing skills. Based on previous research with WS and typical readers we expected that WS individuals would show similar relations to those observed in TD populations, such as FSIQ not being predictive of reading ability whereas specific cognitive skills, including phonemic awareness, phonological decoding, visual-auditory learning and rapid automatic naming were expected to correlate with reading. This has implications for reading intervention in WS, as if the same cognitive correlates are found in WS and in typically developing populations, then this suggests that reading intervention programs used for typical populations may be applicable for those with WS. For the fourth aim, we expected heterogeneity in reading performance at the individual level for both lexical and nonlexical reading, which again has implications for intervention, as it may highlight the need for individualised assessments to be conducted to develop appropriate reading instruction.

Method

Methodological Difficulties in Studying Reading in Atypical Populations

Some studies of reading skills in WS have included a comparison or control group, such as typically developing children (Heinze & Vega, 2008; Laing et al., 2001; Menghini et al., 2004; Temple, 2006) matched on variables such as reading age and verbal mental age (Laing et al. 2001) or just mental age (Menghini et al., 2004, Temple, 2006). Choosing an appropriate comparison group when researching characteristics of individuals with intellectual disability can be problematic (see Brock, 2007; Laing, 2002; Mervis & Klein-Tasman, 2004). If groups are matched on chronological age there can be a lot of variability in intellectual and cognitive skills between the groups. If matched on mental age, this assumes an even profile across various cognitive skills within the groups; however this is not the case for the WS cognitive profile (Laing, 2002; Martens et al., 2008; Porter & Coltheart, 2005). If matched on reading age, there can be significant variability in underlying cognitive abilities (Laing, 2002). In addition, given that reading is a complex skill in itself, it is not clear which particular reading skill should be used to determine reading age. Overall, there is no perfect solution to these methodological issues and differences between studies makes comparison of results difficult. For this study, we decided to use the age at which functional literacy is typically obtained as a benchmark, in that they have achieved a level of reading ability necessary to participate and function reasonably independently within the community. When assessing typically developing readers, they are often compared to the level of reading they should have attained given their age and/or the number of years spent at school. That is, comparisons are made to a level of literacy that should be attained. Since this study investigates how reading ability impacts on everyday functioning, we decided to compare participants to the level of reading that is necessary to perform everyday life tasks.

Participants

Thirty individuals (14 males and 16 females) with a genetically confirmed diagnosis of Williams syndrome (WS) took part in the study (see Porter et al., 2012 for details of genetic confirmation on this sample). The age range of the participants was between 9 years and 39 years with a mean age of 21 years, 4 months. The participants were recruited through the Williams Syndrome Associations of NSW, Victoria and South Australia. Demographic information for the participants is shown in Table 1. The participants' mental age, IQ and verbal ability were assessed using the Woodcock-Johnson III Tests of Cognitive Abilities, Australian Adaptation or WJ III COG (Woodcock et al., 2001). Socio-Economic Status (SES) for each family was based on area of residency (based on postal codes) using the Index of Relative Socio-economic Disadvantage (IRSD). The IRSD is provided online by the Australian Bureau of Statistics (ABS, 2006). The Socio-Economic Index for Areas (SEIFA) percentile score was used, with a lower percentile number reflecting relatively greater disadvantage (lower SES).

Table 1

Demographics for the WS Sample

	Mean (SD) *N* = 30	Range
Chronological age	21y 0m (8y 9m)	9y 5m – 39y 4m
Mental age	7y 7m (1y 9m)	5y 0m – 11y 1m
IQ (GIA)	65.13 (13.67)	25 – 89
Verbal Ability [a]	71.37 (9.93)	38 - 92
SES[b]	58.10 (31.05)	12 - 99

[a] Verbal Ability is significantly higher than IQ (Paired Samples $t(29) = 4.35$, $p < .0005$)

[b] Based on Socio-Economic Index for Areas (SEIFA) percentile score, with a lower percentile number reflecting relatively greater disadvantage (lower SES)

Measures

Castles and Coltheart Reading Test 2 (CC2; Castles et al., 2009). Participants were asked to read aloud words (regular and irregular) and nonwords from the Castles and Coltheart Reading Test 2 (CC2; Castles et al., 2009), a test of single word reading. The CC2 can be used to assess lexical reading (using irregular words) and nonlexical reading (using nonwords). The test contains 120 items made up of 40 regular words (e.g., hand, chicken) and 40 irregular or exception words (e.g., yacht, couple) and 40 pronounceable nonwords (e.g., gop, seldent). Regular and irregular words are matched on frequency, length and grammatical class. Both word lists decrease in frequency as the test progresses. The nonwords increase in length and the complexity of the grapheme-phoneme knowledge required. The test is untimed and the lists are pseudo-randomised. There is a discontinue rule of five consecutive errors that applies separately for each item type. The Cronbach's alpha (indicating internal consistency) has been reported as .85 for the regular subscale, .86 irregular subscale and .94 for the nonword subscale (Moore, Porter, Kohnen, & Castles, 2012). For the purpose of this study, the accuracy score for each subscale was converted to a *z* score using the published norms (Castles et al., 2009).

The normative data for the CC2 had been collected from 1000 typically developing school children (see Castles et al., 2009 for details). The oldest age band (11 years – 11 years 5 months) was used for all participants, apart from three participants whose chronological age fell below this age band (for these participants, aged 9 years 5 months, 10 years 2 months and 10 years 9 months, the age band corresponding to their chronological age was used). While the normative data provided by the CC2 is not appropriate for the majority of participants based on their chronological age, the CC2 was chosen for several reasons. First, the CC2 is the only standardised and normed test that provides separate assessment of lexical and sublexical reading skills which are both important processes in acquiring reading skills. A recent study indicated that mixed lists of regular and irregular words may overestimate word reading abilities compared to separate lists (Moore et al., 2012). In addition, a recent review of nonword tests identified the CC2 as one of the five best nonword reading tests to assess acquisition of sublexical reading skills suitable to an Australian population (Colenbrander, Nickels & Kohnen, 2011). Second, one aim of the current study was to determine whether individuals with WS are 'functionally literate', in that they have achieved a level of reading ability necessary to participate and function reasonably independently within the community. As noted by Wheldall and Watkins (2004), the term functional literacy is difficult to define and there has been little consensus in the literature regarding a level of reading performance at which individuals would be considered functionally literate. Wheldall and Watkins (2004) suggest that reading performance at the end of Year 5 for Australian children as being equivalent to functional literacy level (the chronological age of children in Year 5 is typically from 10 – 11 years). Norming for the CC2 was carried out relatively early in the school year (towards the end of first term or beginning of the second term). Therefore, children in the oldest age band had completed their Year 5 of school and were embarking on Year 6. Thus, using the oldest age band from the CC2 norms was considered the closest to an end of Year 5 level of schooling and representative of a functional level of literacy and was used to evaluate WS participants whose chronological age fell outside of the CC2's norm range.

Vineland Adaptive Behaviour Scales – 2nd Edition (Vineland II; Sparrow et al., 2005). The Survey Interview Form of the Vineland II was administered via a semi-structured interview with the parent or primary carer of each individual to assess their adaptive functioning or everyday living skills. The Vineland II covers three main domains of functioning (Communication, Daily Living Skills, Socialisation) each comprising three subdomains and a fourth domain of Motor skills for individuals aged under seven years. The Motor domain was not administered as all participants were older than seven. The three domain scores are combined to form an Adaptive Behaviour Composite (ABC) score. The two domains of Communication and Daily Living Skills were included in the analysis to examine evidence of how an individual's reading ability compares with their functioning in everyday living. The Socialisation domain was not included as it assesses skills which were thought not likely to be related to reading.

The Communication domain is made up of three subdomains: (1) the *Receptive* subdomain assesses how an individual listens, pays attention and what he/she understands; (2) the *Expressive* subdomain assesses what an individual says, how he/she uses words and sentences to gather and provide information; and (3) the *Written* subdomain assesses what an individual understands about how letters make words and what he/she reads and writes. The three subdomains of the Daily Living Skills domain are: (1) *Personal* skills assess how the individual eats, dresses, practices personal hygiene; (2) the *Domestic* subdomain relates to what household tasks the individual performs; and (3) the *Community* subdomain assesses how the individual understands and uses time and dates, money, telephone, computer and job skills. Internal-consistency reliabilities of the composite ABC and domain scores are generally very high, with most being in the upper .80s to low .90s. Subdomain coefficients are lower and in the .60s through to .80s. Further details of reliability and research supporting the validity of the instrument can be found in the test manual (Sparrow et al., 2005).

Woodcock-Johnson III Tests of Cognitive Abilities - Australian Adaptation (WJ III COG; Woodcock et al., 2001). We chose the WJ III COG battery because Evans, Floyd, McGrew, and Leforgee (2001) recently found that the different subscales of this extensive battery can be used to differentiate reading abilities in typically developing readers. For example, while Fluid Reasoning and Visuo-Spatial abilities were not associated with reading ability, the language subscales showed high associations. All participants in our study completed the extended version of the WJ-III COG (20 subtests). The WJ III COG is based on the Cattell-Horn-Carroll (CHC) theory of cognitive abilities and uses a hierarchical framework consisting of three levels: stratum III is a measure of *g* or general intellectual ability (GIA) similar to FSIQ; stratum II consists of seven broad cognitive (CHC) domains representing the cognitive abilities of: Comprehension-Knowledge, Long-Term Retrieval, Auditory Processing, Visual-Spatial Thinking, Fluid Reasoning, Short-Term Memory, and Processing Speed. Two additional clinical clusters, Working Memory and Phonemic Awareness were included due to their known relationship with reading in typically developing individuals. Stratum I consists of the narrow or individual cognitive abilities. The Comprehension-Knowledge domain on the WJ III COG is also used as the measure of Verbal Ability, assessing language based acquired knowledge. The WJ III COG has high reliability and validity as outlined in the WJ III COG technical manual (McGrew & Woodcock, 2001). The median reliabilities for the clusters are .90 or higher and nearly all individual subtests have median reliability coefficients of .80 or higher.

Following is a brief description of the two individual subtests (stratum I) which contribute to each CHC and Clinical Cluster (stratum II) of the WJ III COG used in the analysis (Mather & Woodcock, 2001; Schrank, Flanagan, Woodcock & Mascolo, 2002).

Comprehension - Knowledge (*Gc*): *Verbal Comprehension (Subtest 1)* includes picture vocabulary (naming pictures), synonyms and antonyms (providing the synonym or antonym for a given word), and verbal reasoning (completing an analogy with an appropriate word). *General Information (Subtest 11)* asks questions of general verbal knowledge “What would you do with…” and “Where would you find…(an object)”.

Long-Term Retrieval (*Glr*): *Visual-Auditory Learning (Subtest 2)* is a measure of associative learning. Individuals are asked to learn and recall pictographic representations of words. *Retrieval Fluency (Subtest 12)* assesses verbal fluency for three specified categories (e.g., name as many animals as you can in 60 seconds).

Visual-Spatial Thinking (*Gv*): *Spatial Relations (Subtest 3)* requires visualisation of spatial-relationships. Individuals are asked to identify pieces that form a target shape. *Picture Recognition (Subtest 13)* is a measure of visual memory of objects or pictures. Individuals are asked to recognise previously presented pictures from an array containing distractors.

Auditory Processing (*Ga*): *Sound Blending (Subtest 4)* requires listening to a series of syllables or phonemes presented by the examiner and then blend the sounds into a word. *Auditory Attention (Subtest 14)* assesses speech-sound discrimination. The individual has to listen to individual words from an audio recording amid increasingly distorting background noise.

Fluid Reasoning (*Gf*): *Concept Formation (Subtest 5)* involves inductive logic. The individual has to examine a stimulus set and formulate a rule that applies to the item. *Analysis-Synthesis (Subtest 15)* involves sequential reasoning. The individual has to solve a series of increasingly complex puzzles

Processing Speed (*Gs*): *Visual Matching (Subtest 6)* requires individuals to locate and circle two identical numbers in a row of six, with a 3 minute time limit. *Decision Speed (Subtest 16)* assesses ability to make conceptual decisions quickly. They are asked to examine a row of objects and mark the two pictures which go together conceptually, with a 3 minute time limit.

Short-Term Memory (*Gsm*): *Numbers Reversed (Subtest 7)* asks individuals to initially hold a series of numbers and then perform a mental operation (i.e., reverse the sequence). *Memory for Words (Subtest 17)* asks individuals to repeat lists of unrelated words in the correct sequence.

Phonemic Awareness: *Sound Blending* (as described in Auditory Processing above). *Incomplete Words (Subtest 8)* asks the participant to listen to a word that has one or more phonemes missing and identify the complete word.

Working Memory: *Numbers Reversed* (as described in Short-Term Memory above). *Auditory Working Memory (Subtest 9)* asks the individual to listen to a series of digits and words and reorder the information by repeating back first the objects and then the digits.

One additional subtest, *Rapid Picture Naming (Subtest 18)* from the WJ III COG was included as a test of rapid automatic naming (RAN) which has been found to be associated with word reading ability (Arnell et al., 2009). It requires the rapid naming of pictured common objects within 2 minutes.

Procedure

Administration. Each participant was tested individually either at home or in clinical rooms at Macquarie University. Due to the length of the test battery, individuals were provided with rest breaks as required and for some individuals, testing was completed across two separate sessions. Standardised tests were administered according to the examiner's manual. An audio recording is available for presentation of subtests 4, 7, 8, 9, 14 and 17 but can be presented orally if subjects have difficulty paying attention or dislike wearing headphones (Schrank et al., 2002). To maintain participant's attention, all subtests were presented orally, apart from subtest 14 for which the audio recording is essential as it provides simultaneous presentation of oral language with increasing louder background noise. The semi-structured interview of the Vineland II was conducted with the primary carer (typically parent) either on the same day as cognitive testing (during the rest break) or at a later date via telephone interview. The CC2, WJ III COG and Vineland II were administered by the first author of this paper; a trained intern clinical neuropsychologist. Administration and scoring was overseen by the third author, a fully registered clinical neuropsychologist.

Scoring. All tests were hand scored using standardised instructions outlined in the administration manuals (Castles et al., 2009; Mather & Woodcock, 2001, Sparrow et al., 2005) and the WJ III and Vineland II scoring was checked using relevant computer scoring programs. Computer scoring of the WJ III COG was completed using the WJ III Normative Update (NU) Compuscore and Profiles Program – Australian Adaptation, Version 1.0 (Schrank & Woodcock, 2009). The Vineland-II was scored using the Vineland II ASSIST (2008) Scoring and Reporting computer program, Version 1.2.

Statistical Analyses. Prior to analysis, scores were examined for missing values, normality and outliers. Where multiple post-hoc comparisons were conducted, a Bonferroni adjustment was made according to the number of comparisons. Where multiple correlations were conducted, the alpha level of significance was adjusted to .01 to minimise the chance of committing a Type I error. Statistical analyses were conducted using IBM SPSS Statistics Standard Grad Pack 21 computing software.

Results

As participants in the current study underwent a full genetic work-up, with some genetic variability observed within the cohort (see Porter et al. 2012 for further details), the results start with an analysis to determine whether it was appropriate to retain participants with an atypical WS deletion in the study. The remaining results are then organised into four parts: (1) CC2 group profile of reading; (2) the relationship between reading and adaptive functioning; (3) the relationship between reading and cognitive functioning (including IQ and more specific cognitive abilities) as measured by the WJ III COG; and, (4) evidence of different reading profiles based upon the predictions of the Dual Route model.

Genetic variability and reading ability.

Based on full genotyping using genome array screening (Porter et al., 2012), 22 individuals in the current sample were identified as having the typical ~1.6Mb WS deletion (Schubert, 2009) and eight individuals were identified as having an atypical WS deletion (a larger 1.8Mb deletion, involving three extra genes to the ~1.6Mb deletion). To determine whether there was a significant difference in reading abilities between the individuals with the typical deletion and those with an atypical deletion, individual matched pairs were created. Each individual with an atypical deletion was matched to a participant with a typical deletion on the basis of their gender, chronological age (CA), mental age (MA), and intellectual functioning GIA (IQ). Paired sample t-tests confirmed there was no significant difference between the matched pairs for CA [$t(7) = -1.14$, $p = .290$, Typical Mean = 22.42 years (s.d. = 8.40 years), Atypical Mean = 24.31 years (s.d. = 7.94)]. There was also no significant difference for MA [$t(7) = -.199$, $p = .848$, Typical Mean = 7.77 years (s.d. = 2.01), Atypical Mean = 7.99 years (s.d. = 2.06)] or GIA [$t(7) = -1.39$, $p = .207$, Typical Mean = 66.88 (s.d. = 18.87), Atypical Mean = 67.38 (s.d. = 15.91)].

On the CC2, the two groups were found to be highly correlated for each reading list (regular words $r = .755$, $p = .030$; irregular words $r = .691$, $p = .058$; nonwords $r = .589$, $p = .125$). Using paired t-tests, there was no evidence of a significant difference in reading ability for each word type between those with a typical or atypical deletion [regular words, $t(7) = 1.96$, $p = .09$; irregular words, $t(7) = 1.45$, $p = .19$; nonwords, $t(7)$

= 1.59, p = .15]. Based on these results, which indicated similar reading patterns across WS patients with the typical and 1.8Mb deletion, it was decided to retain the eight participants with an atypical deletion in the current study.

CC2 Reading Profile

Reading results for the group of 30 participants on the subscales of the CC2 are presented in Table 2. The results show that reading ability was highly variable between the participants. Some participants could not read any words or nonwords correctly, while others made few errors on the CC2 reading test. The sample included two participants who could not read any words or nonwords at all, and were classified as non-readers. Levy et al., 2003 questioned whether individuals who cannot read at all should be excluded in results of studies investigating reading ability. They described Laing et al.'s (2001) study as having methodological problems due to their inclusion of three non-readers within their group of 15 participants, as this would have had the effect of lowering the overall reading age of the group and, therefore, impacted on the age of the matched control group resulting in children as young as five years being included as controls. Also, they noted that Laing et al. did not state whether the non-readers were the youngest in the group and possibly in the early stages of learning to read. The current study did not use a matched control group, so inclusion of the two non-readers was not an issue in relation to matched controls[8]. In fact, deleting these individuals would reduce variance in our investigations of the relationships between reading and IQ or adaptive functioning.

Table 2

Group Performance on the CC2 (Means, Standard Deviations, Range and, Age Equivalents for Scores on Regular, Irregular and Nonword Reading Subscales)

	Raw Score[a]	z-score[b]	
	Mean (SD)	Mean (SD)	Mean AE[c] (SD)

[8] The non-readers in this study were both females and similarly aged (32y 10m and 33y and 7m) and therefore well past the age of reading acquisition. One had an atypical WS deletion and one had the common WS deletion. They had a similar level of intellectual functioning and did not have the lowest IQ scores in the group (GIA or IQ was 67 and 65 respectively).

CC2 Subscales	Range	Range	Range
Regular words	24.33 (13.79)	-1.28 (1.27)	8y 1m (1y 9m)
	0 – 39	-3.09 to 1.10	< 6y 0m to > 11y 5m
Irregular words	16.33 (8.80)	-1.16 (1.10)	8y 5m (1y 7m)
	0 – 29	-3.09 to 0.94	< 6y 0m to 10y 8m
Nonwords	12.83 (12.49)	-1.87 (0.97)	7y 3m (1y 4m)
	0 – 34	-3.09 to -0.29	< 6y 0m to 10y 2m

[a]Accuracy raw score is the number of correct items out of 40 for each subtest

[b]z-scores have a mean of 0 and standard deviation of 1

[c]AE= Age Equivalent score. Youngest age on norm tables is 6y 0m and oldest age is 11y 5m

The relationship between the reading of regular, irregular, and nonwords (as measured by the CC2) was investigated using Pearson correlation coefficients. Preliminary analyses were performed to confirm no violation of the assumptions of normality, linearity and homoscedasticity. There were very strong positive correlations between all subscales of the CC2: Regular and Irregular reading, r = .812; Regular and Nonword reading, r = .923; Irregular and Nonword reading, r = .774 (for all correlations, p < .0005).

A one-way repeated measures ANOVA was conducted to further compare scores on the subscales of the CC2. The means and standard deviations are presented in Table 2. There was a significant effect for reading subscale, Wilks's Lambda = .358, $F(2,28) = 25.056$, $p < .0005$, multivariate $\eta_p^2 = .642$. Follow-up Bonferroni adjusted comparisons (.05/3) revealed that both regular and irregular word reading scores were significantly higher than nonword reading scores ($p < .0005$). There was no significant difference between regular and irregular word reading ($p = 1.00$), indicating no regularity effect (better reading of regular versus irregular words) for the group.

Reading Ability and Adaptive Functioning

Descriptive statistics for the overall adaptive functioning composite (ABC) and for Communication and Daily Living Skills domains and their respective subdomains on the Vineland II are presented in Table 3. Overall, as a group, the participants' adaptive functioning was Low, however, as can be seen from the range of scores in Table 3, there was considerable variability.

Table 3

Descriptive Statistics for Group Performance on the Vineland II

Vineland II Subdomain	Mean (SD) *n* = 29	Range
Adaptive Behaviour Composite (ABC)[a]	**55.07 (14.43)**	**20 - 84**
Communication Skills[a]	**51.76 (20.22)**	**21 - 82**
Receptive[b]	7.65 (2.99)	1 – 15
Expressive[b]	7.83 (2.32)	2 – 13
Written[b]	8.00 (2.48)	2 – 14
Daily Living Skills[a]	**57.76 (13.89)**	**31 – 93**
Personal[b]	7.97 (2.81)	3 - 16
Domestic[b]	7.31 (3.26)	1 - 16
Community[b]	7.17 (2.83)	1 – 12

Note. [a]ABC and adaptive domain standard scores have a mean of 100 (*SD* = 15), range 20-160. Scores ≤ 70 are impaired. [b]Subdomain *v*-scale scores with a mean of 15 (*SD* = 3), range 1-24. Scores ≤ 9 are impaired. For domains and subdomains, a higher score indicates better functioning for the individual. Domain scores more than two standard deviations below the population mean (standard score < 70) are considered impaired level. For subdomains, scores equal to, or less than 9 are considered impaired.

The relationships between regular, irregular and nonword reading ability on the CC2 and adaptive functioning skills on the Vineland II were investigated using Pearson correlations. These correlations are presented in Table 4.

Table 4

Correlations between Adaptive Functions on the Vineland II and Subscales of the CC2

Vineland II	CC2 Subscales		
Domains & Subdomains	Regular Words	Irregular Words	Nonwords
ABC	.359	.486*	.506**
Communication	.251	.437	.446*
Receptive	.226	.447	.362
Expressive	.467*	.518*	.585*
Written	.506**	***.631****	***.597****
Daily Living Skills	.415	.503**	.493*
Personal	.281	.387	.315
Domestic	.189	.261	.232
Community	***.670****	***.715****	***.714****

Note. *$p \leq .01$ **$p \leq .005$, ***Bold italics*** = correlations which remain significant (all at $p \leq .005$) when intellectual functioning (IQ) is held constant.

As seen in Table 4, an individual's functional reading ability was related to aspects of their adaptive functioning. Specifically, within the Communication domain, reading ability for all CC2 subscales was found to be associated with an individual's use of spoken (Expressive) and written language skills (Written subdomain assesses what an individual reads and writes). The other significant correlations were between each of the three subscales of the CC2 and the Community skills subdomain. This indicates that higher functional reading ability is related to having better skills in everyday tasks involving concepts such as time, money, telephone, computer, etc. When IQ is held constant, reading skills were still significantly associated with Written and Community skills.

Relationship between Reading and Intellectual and Cognitive Abilities

Pearson correlations were conducted to examine the relationship between reading ability on the subscales of the CC2 (regular, irregular and nonwords) with chronological age, mental age, GIA (IQ) and CHC cognitive factors (Stratum II) of the WJ III COG. Results are presented in Table 5. Although significance level set at .01 for multiple comparisons, correlations significant at $p < .05$ are also indicated in Table 5 for interests sake.

Table 5

Correlations Between Reading Subtypes on the CC2, Chronological Age, Mental Age, GIA (IQ), and WJ III COG Cognitive Domains

	Regular Word	Irregular Word	Nonword
Chronological Age	-.126	-.147	-.288
Mental Age	.665***	.651***	.698***
General Intellectual Ability (IQ)	.551**	.538**	.587**
Comprehension-Knowledge (*Gc)*.	.381*	.425*	.451*
Long-Term Retrieval (*Glr*)	.491*	**.523****	**.558****
Visual-Spatial Thinking (*Gv*)	.260	.214	.346
Auditory Processing (*Ga)*	**.672*****	**.647*****	**.634*****
Fluid Reasoning (*Gf)*	.447*	.441*	.431*
Processing Speed (*Gs*)	.296	.355	.400*
Short-term Memory (*Gsm)*	**.598*****	**.572****	**.661*****
Phonemic Awareness (PA)	**.732*****	**.650*****	**.669*****

Working Memory (WM)	.469*	.449*	**.563****

Note. *$p \leq .05$ **$p \leq .01$** ***$p \leq .0005$**

Correlations between CC2 reading subscales and chronological age were not statistically significant. Reading ability was, however, significantly and positively correlated with mental age and global intellectual ability (GIA). Correlation coefficients were similar across regular, irregular and nonwords, but nonwords seemed to be particularly highly correlated with mental age and GIA (IQ).

Three cognitive domains were found to be highly correlated with all CC2 reading subscales; Auditory Processing (*Ga),* Short-Term Memory (*Gsm*) and Phonemic Awareness. With the adjusted alpha level of .01 for multiple comparisons, Long-Term Retrieval (*Glr)* was significantly correlated with irregular and nonword reading and Working Memory with nonword reading. All of these WJ III COG domains have also been shown to be related to reading ability for typically developing children and adolescents (Evans et al., 2001). Two additional domains which were found to be correlated with reading ability in typically developing individuals, Comprehension-Knowledge (*Gc*) and Processing Speed (*Gs*), were not significantly related to reading ability for the WS group once the alpha level was adjusted for multiple comparisons. Verbal Comprehension is combined with the General Information subtest to form the Comprehension-Knowledge (*Gc*) factor. For typically developing readers, Comprehension-Knowledge from the WJ III COG has been found to be the largest predictor of reading achievement (Evans et al., 2001). With the known variability of the WS cognitive profile, the use of broad domain scores may mask some relative strengths and weaknesses. For example, receptive vocabulary is known to be a relative strength in WS (Mervis, 2009) and is included as part of the Verbal Comprehension subtest of the WJ III COG. Therefore, correlations between individual subtests (narrow abilities) which comprise each domain and subscales of reading were also investigated and are shown in Table 6.

Table 6

Correlations between Reading Ability on the CC2 and WJ III COG Subtests

WJ III Domains:	*Gc*	*Gc*	*Glr*	*Glr*	*Gv*	*Gv*		*Ga*	*Ga*	*Gf*	*Gf*
WJ III Subtests:	1	11	2	12	3	13		4	14	5	15
CC2 Subscales											
Regular Words	**.506****	.173	**.611*****	.283	.467*	-.137		**.733*****	.024	.342	**.514****
Irregular Words	**.551****	.201	**.598*****	.386*	.397*	-.087		**.616*****	.239	.329	**.563****
Nonwords	**.562****	.231	**.626*****	.390*	.490*	-.011		**.666*****	.106	.295	**.564****
Domain level	*Gs*	*Gs*	*Gsm*	*Gsm*	PA		PA	WM	WM	Test 10	Test 18
Subtest Level	6	16	7	17	4		8	7	9	(n=17)	
CC2 Subscales											
Regular Words	.345	.287	**.597*****	.462*	**.733*****		**.538****	**.597*****	.094	.014	.142
Irregular Words	.427*	.343	**.602*****	.426*	**.616*****		**.567****	**.602*****	.106	.138	.292
Nonwords	**.510****	.354	**.700*****	.484*	**.666*****		**.521****	**.700*****	.188	.108	.231

Note. **p* ≤ .05, *******p* **≤ .01**, ********p* **≤ .0005**; *Comprehension-Knowledge (Gc):* 1 = Verbal Comprehension, 11 = General Information; *Long Term Retrieval (Glr)*: 2 = Visual-Auditory Learning; 12 = Retrieval Fluency; *Visual-Spatial Thinking (Gv*): 3 = Spatial Relations 13 = Picture Recognition; *Auditory Processing (Ga):* 4 = Sound Blending; 14 = Auditory Attention; *Fluid Reasoning (Gf)* = 5 = Concept Formation; 15 = Analysis-Synthesis; *Processing Speed (Gs):* 6 = Visual Matching, 16 = Decision Speed; *Short-Term Memory (Gsm):* 7 = Numbers Reversed; 17 = Memory for Words; *Phonemic Awareness (PA):* 4 = Sound Blending , 8 = Incomplete Words; *Working Memory (WM*): 7 = Numbers Reversed; 9 = Auditory Working Memory.

Additional Subtests: Test 10 = Visual-Auditory Learning Delayed (n= 17); Test 18 = Rapid Picture Naming

The subtests which were found to be most strongly associated with all reading subscales were: Verbal Comprehension (which contributes to the Comprehension-Knowledge *Gc* WJ-III COG domain), Visual-Auditory Learning, (which contributes to the Long Term Retrieval *Glr* domain), Sound Blending (which contributes to both the Auditory Processing *Ga* domain and the Phonemic Awareness domain), Analysis-Synthesis (which contributes to the Fluid Reasoning *Gf* domain); Numbers Reversed (part of both the Working Memory and the Short-Term Memory *Gsm* domains) and Incomplete Words (Phonemic Awareness domain). Visual matching (part of the Processing Speed *Gs* domain) was significantly related to nonword reading. It is interesting to note that when the two subtests which contribute towards each broad cluster are examined, only one of the narrow abilities which make up that cluster was found to be associated with reading. This suggests that it is necessary to consider the individual subtests rather than just the broad cognitive domains, as domain scores may hide some of the variability and strengths and weaknesses for individuals with WS.

Phonemic Awareness was the only broad domain in which both contributing subtests were found to be associated with reading (Sound Blending and Incomplete Words). Cognitive areas which were not related to word reading included: General Information, Retrieval Fluency, Spatial Relations, Picture Recognition, Auditory Attention, Concept Formation, the delayed version of Visual-Auditory Learning, (only 17 out of the 30 participants completed this task due to error cut-off rules), Decision Speed, Memory for Words, Auditory Working Memory, and Rapid Picture Naming.

Cognitive Predictors of Reading Ability

Standard multiple regression was used to assess the ability of three cognitive skills (associative learning, phonemic awareness, working memory) to predict reading ability, first for nonword reading and then for irregular word reading. Due to the small sample size, the analysis was restricted to three predictors. Subtest 2, Visual-Auditory Learning, was the measure of associative learning; Subtest 4, Sound Blending, was the measure of phonemic awareness; and Subtest 7, Numbers Reversed, was used as the measure of working memory. The three abilities were chosen as they have been found to be predictive of reading ability in typically developing individuals and were identified as being correlated with reading for these WS participants. Preliminary analyses were conducted to ensure no violation of the assumptions of normality, linearity, multicollinearity and homoscedasticity. The model, which included measures of associative learning, working memory and phonemic awareness, explains 58.9% of the variance in nonword reading, $F(3,26) = 14.83$, $p < .0005$. Of the three variables, working memory made the largest unique contribution (beta = .387, $p = .02$), while the contributions made by associative learning (beta = .272, $p = .076$) and phonemic awareness (beta = .285, $p = .084$) were not significant. Using the same three predictors for irregular word reading, 46.7% of the variance was explained, $F(3,26) = 9.46$, $p < .0005$. None of the variables made a unique contribution (associative learning, beta = .302, $p = .084$; sound blending beta = .288, $p = .123$; numbers reversed beta = .272, $p = .137$). Due to the small sample size, statistical analysis of these reading abilities by subgroup was not possible, however means and standard deviations for each subtest of the WJ III COG for the group as a whole and by subgroups is provided in Table 7.

Is Reading On Par with IQ?

At an individual level, the profile of each participant's reading ability (z-score) was compared to the z-score of intellectual ability (the scoring program of the WJ III COG provides z-scores as well as standard scores for GIA). A difference of 1.5 points between reading ability z-score and intellectual ability z-score (i.e., a *z*-score difference of either $\leq$ - 1.5 or $\geq$ +1.5) was used to indicate a significant difference between reading ability and intellectual functioning. From the 28 readers, the majority displayed no significant difference between intellectual ability and reading ability for each reading subscale, as the difference between their GIA z-score and reading z-score was between the range of -1.49 through to +1.49 (regular words, 68% , $n = 19$; irregular words 61%, $n = 17$; nonwords, 89%, $n = 25$). However, there were some individuals who showed significantly higher reading scores compared to their GIA (regular words, 32%, $n = 9$, of which, two had the atypical deletion; irregular words, 39%, $n = 11$ of which two had the atypical deletion; nonwords, 11%, $n = 3$, all typical deletion). No individuals showed the reverse pattern of a significantly higher GIA z-score ($\geq$ 1.5) compared to their reading z-scores.

In summary, for the majority of individuals, IQ was consistent with their reading ability, whereas 32% (regular words), 39% (irregular words) and 11% (nonwords) of individuals showed a significantly higher reading score than IQ. No individuals showed the reverse pattern of significantly higher IQ than reading.

Reading Subgroups

The majority of previous studies have found that individuals with WS tend to have poorer nonlexical than lexical reading skills. However, the reverse pattern has also been found. We wanted to investigate what type (s) of reading profile(s) would be present in our sample. We developed a classification system based on the Dual Route theory. According to the Dual Route theory, irregular words can only be read via the lexical route while pronounceable nonwords can only be read correctly via the nonlexical route. Difference in performance between these two item types was of primary interest to determine whether any participants could be classified as relying on their lexical reading route *Lexical Readers* (poorer nonword than irregular word reading). We were also interested in *Nonlexial Readers* who show over-reliance on nonlexical reading (the reverse pattern of poor irregular word reading and better nonword reading) or *Mixed Readers* (poor performance on both word types). Individuals who could read both word types at a functional level (within 1 s.d. of the standardization sample mean and at Grade 5 level) were classified as *Functional Readers.*

We used similar cut-offs to those described in McArthur et al. (2013) whereby the z-score performance for the subsets of word types of interest on the CC2 (i.e., irregular words and nonwords[9]) for each individual was analysed. Functional performance was defined as falling within the average range or within one standard deviation of the mean (z scores between -1.0 and +1.0), while for inferior performance, a cut-off z-score of ≤ -1.3 was used, which identifies the lowest 10% of the normative population. Using this criterion, four different reading subgroups were identified.

1. Functional Readers: 8 participants (27%) scored within the average range (*above* the level of -1.00 z-score) on both irregular word and nonword reading subscales.
2. Lexical Readers: 6 participants (20%) met the criteria for a sublexical impairment, with their lexical (irregular word) reading score ≥ -1.0, while their sublexical (nonword reading) was ≤ -1.30.[10]

[9] Since regular words can be read by either the lexical or nonlexical route, they were not relevant for this analysis.

[10] Note that two individuals (Identification numbers 13 and 15 in Table 7) did not fit the criteria exactly for the Lexical Readers subgroup, but this group was the closest fit that could be found to place them in.

3. Nonlexical Readers: No participants showed this pattern where nonword reading is within the average range, while irregular word reading is inferior (z-score ≤ -1.30).
4. Mixed Readers: 16 participants (53%) were classified as reading below a functional level with their z-scores for both irregular words and nonwords < -1.0.

In addition to the profiles based on McArthur et al. (2013) for this sample, it seemed prudent to make further differentiations in the mixed group:

i) Nonfunctional Readers: Nine of the 17 participants in the 'Mixed readers' category were classified as reading below a functional level with their z-scores for both irregular words and nonwords < -1.0.

ii) Nondecoders: Five individuals were classified as nondecoders, as they were unable to read any of the nonwords. However, they could read either one or more regular or irregular words.

iii) Nonreaders: Two readers could not read any words on all three subscales.

Table 7 provides descriptive statistics, for each reading subgroup, as well as its individual members. Details on gender, genetic deletion type (typical or atypical), chronological age, mental age, GIA (IQ) score, Verbal Ability, and irregular and nonword z-scores are detailed for each participant separately. As can be seen in Table 7, the eight individuals with an atypical deletion were evenly distributed throughout the reading groups, with either one or two individuals with an atypical deletion being included in each group. Mental age, GIA (IQ) score and Verbal Ability were determined by performance on the WJ III COG. Figure 1 shows the patterns of means for each reading group across the three reading subscales of the CC2. The horizontal line on Figure 1 corresponds to a z-score of -1.0 (the cut off for functional reading performance). Figure 1 highlights the different reading performance across all reading subscales for the Functional and Lexical groups and the three subgroups within the Mixed group.

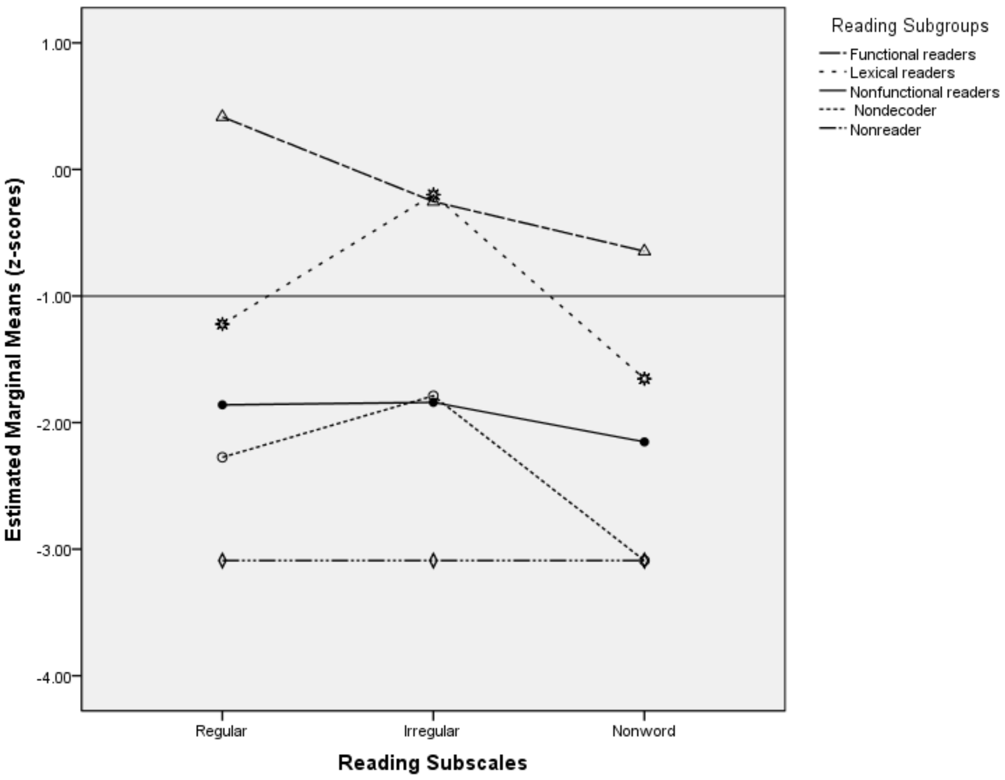

Figure 1. Each reading subgroups' performance across reading subscales.

A mixed between-within subjects analysis of variance (ANOVA) was conducted to assess the difference between reading subgroups on the reading subscales of interest (irregular words and nonwords). Univariate results are presented, as Mauchly's test indicated that the assumption of sphericity was met [Mauchly's W = 1.0, $\chi^2 = .0$, Greenhouse-Geisser and Huynh-Feldt ($\varepsilon = 1.0$)]. As the groups were based on the differences between reading scores on different reading subscales, it is not surprising that a significant interaction between Reading Subscales and Reading Subgroups was found [$F(4,25) = 8.52$, $p < .0005$, $\eta_p^2 = .577$].

Follow-up tests of simple effects were conducted to determine whether there was a significant difference between the two reading subscales of interest (irregular vs nonwords) for the four reading subgroups (Functional, Lexical, Nonfunctional and Nondecoders). Due to the number of comparisons, the alpha level was adjusted (.05/4). A highly significant difference, with higher mean reading score for irregular words compared to nonwords, was confirmed for the Lexical Reading group and the Nondecoders (both *p* values < .0005). There was no significant difference in irregular and nonword reading for the Functional and Nonfunctional reading groups ($p = .034$

and *p* = .070, respectively). For the Functional reading group, tests of simple effects revealed that their regular word reading ability was significantly higher than both their irregular word and nonword reading abilities (both *p* values < .0005). This pattern suggests that for these individuals, both the lexical and nonlexical routes were utilised to assist with regular word reading, resulting in significantly higher scores for regular word reading. This pattern is also observed in typically developing readers (Castles et al., 2009). Overall, the analyses confirm our groupings.

A mixed between-within subjects analysis of variance with GIA (IQ) included as a covariate (ANCOVA) was then conducted to test the interaction between Reading Subscale and GIA along with the interaction between Reading Subscale and Reading Subgroup. This enabled a test of whether the apparent interaction between Reading Subscale and Reading Subgroup could be accounted for by GIA. The interaction between Reading Subscale and GIA was not significant, $F(2,48) = 1.924$, $p = .157$, $\eta_p^2 = .074$. However, Reading Subscale by Reading Subgroup remained significant $F(8,48) = 9.269$, $p < .0005$, $\eta_p^2 = .607$. This demonstrates that the significant interaction between Subgroup by Word Type cannot be accounted for by GIA.

Table 7

Descriptive Statistics for each Reading Subgroup

ID	M/F	DEL	CA	MA	GIA	Verbal Ability	Irreg z-score	Nonwords z-score
Functional Reading Subgroup								
27	F	T	15y 2m	9y 0m	74	75	0.94	-0.29
20	M	A	28y 6m	9y 11m	86	85	-0.15	-0.29
7	F	T	29y 6m	10y 6m	89	92	-0.15	-0.67
26	M	T	18y 4m	9y 9m	74	66	-0.24	-0.65
6	M	T	11y 9m	7y 0m	66	81	-0.24	-0.74
5	M	T	22 y2m	8y 0m	62	59	-0.45	-0.65
22	M	T	14y 8m	7y 7m	65	72	-0.87	-0.94
41	F	T	32y 9m	7y 7m	78	82	-0.87	-0.94

Mean			21y 7m	8y 8m	74.25	76.50	-0.25	-0.65
(SD)			7y 10m	1y 3m	9.81	10.70	0.57	0.25
Lexical Reading Subgroup								
15	F	A	24y 7m	11y 1m	81	82	0.74	-1.20
4	F	T	13y 8m	7y 10m	69	78	0.90	-1.60
35	M	T	20y 1m	8y 8m	66	73	-0.24	-2.28
24	F	T	10y 2m	6y 4m	62	74	-0.70	-1.40
28	F	T	13y 0m	6y 5m	56	66	-0.87	-1.67
13	F	A	18y 4m	9y 0m	69	78	-1.02	-1.77
Mean			16y 7m	8y 2m	67.17	75.17	-0.20	-1.65
(SD)			5y 3m	1y 9m	8.38	5.53	0.83	0.37
Mixed Readers								
i) Nonfunctional Reading Subgroup								
12	M	T	18y 9m	10y 10m	79	75	-1.10	-1.20
25	F	A	16y 9m	8y 10m	70	74	-1.20	-1.64
10	F	T	15y 9m	8y 7m	70	66	-1.30	-1.62
42	M	T	17y 6m	9y 4m	72	66	-1.77	-2.02
11	M	T	19y 0m	7y 7m	61	69	-1.93	-2.69
3	M	T	9y 5m	5y 6m	50	62	-2.17	-2.69
21	F	T	10y 9m	5y 8m	48	63	-2.22	-2.13
23	F	T	14y 5m	6y 33m	52	73	-2.29	-2.69
17	F	A	14y 5m	7y 4m	63	76	-2.58	-2.69
Mean			15y 2m	7y 9m	62.78	69.33	-1.84	-2.15
(SD)			3y 4m	1y 9m	10.92	5.33	0.53	0.57

ii) Nondecoders

16	F	A	36y 3m	6y 10m	72	73	-1.10	-3.09
9	M	T	39y 4m	5y 10m	63	64	-1.20	-3.09
8	M	T	37y 4m	6y 3m	67	75	-2.12	-3.09
32	M	T	20y 6m	5y 0m	25	38	-2.23	-3.09
34	F	A	22y 1m	5y 4m	33	59	-2.29	-3.09
Mean			31y 1m	5y 10m	52.00	61.80	-1.78	-3.09
(SD)			9y 0m	0y 8m	21.42	14.82	0.59	0.0

iii) Nonreaders

14	F	T	32y 10m	5y 7m	67	74	-3.09	-3.09
33	F	A	33y 7m	5y 7m	65	71	-3.09	-3.09
Mean			33y 2m	5y 7m	66	72.50	-3.09	-3.09
(SD)			0y 6m	0y 0m	1.41	2.12	0.0	0.0

Note. ID= Identification number; M/F = Male or Female; DEL= Deletion size (either A=Atypical or T=Typical deletion); CA = Chronological Age in years, months; MA = Mental age in years, months; GIA = General Intellectual Ability (FSIQ); Verbal Ability = Verbal standard score. Both GIA and Verbal Ability are standardised scores with a mean of 100, standard deviation of 15. GIA, Verbal Ability and MA determined by the WJ III COG; Irreg z-score = CC2 Irregular subscale z-score; Nonwords = CC2 Nonword subscale z-score.

Table 8 provides means (and standard deviations for each reading subgroup on each WJ III COG subtest (narrow or Stratum I abilities) and highlights that the subgroups demonstrated similar levels of cognitive abilities across a range of tasks. One way ANOVAs were conducted to assess whether there was a significant difference between the reading subgroups on each cognitive subtest and overall GIA (IQ). Due to the large number of analyses and the small sample, the overall alpha level of .01 was used to determine significance. The difference between the reading subgroups for GIA (IQ) was not significant, $F(4,25) = 2.63$, $p = .059$. As indicated in Table 8, significant differences between the reading subgroups were identified on Sound Blending (phonemic awareness, $F(4,25) = 5.59$, $p < .005$) and Numbers Reversed (working memory, $F(4,25) = 5.34$, $p < .005$). Two subtests approached significance - Verbal Comprehension, ($F(4,25) = 3.64$, $p = .018$) and Visual Auditory Learning (paired associative learning, $F(4, 25) = 3.42$, $p = .023$). Due to the small sample sizes comprising the subgroups, follow-up analyses were not conducted. However, Table 8 indicates, for example, that ND and NR subgroups performed lowest on Sound Blending and on Numbers Reversed. The ND group also seemed to perform lowest on Verbal Comprehension and NF, ND and NR groups all performed lowest on Visual Auditory Learning.

Table 8

Means and (Standard Deviations) for Individual Subtests of the WJ III COG for the Whole Group and for each Reading Subgroup

	Group	Reading Subgroups				
		F	L	NF	ND	NR
WJ III COG Subtests	*N*=30	*n*=8	*n*=6	*n*=9	*n*=5	*n*=2
1. Verbal Comprehension	73.03	80.63	74.83	70.56	63.40	72.50 [a]
	(9.68)	(8.26)	(7.86)	(6.37)	(12.28)	(2.12)
2. Visual-Auditory Learning	63.90	74.75	71.17	56.89	52.40	59.00 [b]
	(15.32)	(8.90)	(15.60)	(16.99)	(7.45)	(9.90)
3. Spatial Relations	76.00	81.88	76.17	74.44	70.00	74.00
	(8.26)	(6.66)	(10.19)	(7.13)	(7.52)	(5.66)
4. Sound Blending	100.67	115.25	104.17	98.44	84.40	82.50 [c]
	(16.67)	(10.04)	(18.02)	(12.31)	(13.24)	(3.54)
5. Concept Formation	70.40	74.00	69.83	71.56	65.20	65.50
	(14.24)	(9.44)	(19.71)	(17.89)	(10.45)	(3.54)
6. Visual Matching	53.50	57.88	59.67	55.00	38.20	49.00
	(15.94)	(13.50)	(16.73)	(16.37)	(15.07)	(5.66)
7. Numbers Reversed	65.13	78.00	71.33	66.56	42.60	45.00 [d]
	(19.20)	(8.16)	(13.15)	(10.90)	(29.20)	(8.48)
8. Incomplete Words	96.53	101.88	101.67	96.22	86.40	86.50
	(13.14)	(17.12)	(10.48)	(8.41)	(13.45)	(3.54)
9. Auditory Working Memory	71.27	73.38	73.67	74.11	58.00	76.00
	(15.50)	(9.80)	(13.40)	(13.08)	(26.75)	(9.90)
10. Visual-Auditory Delay[*]	60.41	56.67	68.80	57.17	-	-
	(12.96)	(11.59)	(10.96)	(14.40)		

11. General Information	73.60	74.50	78.33	73.78	65.80	74.50
	(11.46)	(14.09)	(7.09)	(7.29)	(17.98)	(0.71)
12. Retrieval Fluency	74.80	79.63	80.00	77.67	57.20	71.00
	(17.19)	(11.88)	(15.01)	(6.22)	(32.21)	(0.0)
13. Picture Recognition	85.30	84.88	85.50	86.22	84.20	85.00
	(5.92)	(4.09)	(6.77)	(8.07)	(5.59)	2.83
14. Auditory Attention	79.00	75.50	87.83	80.89	73.40	72.00
	(17.46)	(25.18)	(13.20)	(9.14)	(20.77)	(16.97)
15. Analysis Synthesis	71.50	77.63	74.67	70.00	62.00	68.00
	(13.98)	(9.68)	(6.65)	(18.99)	(16.14)	(2.83)
16. Decision Speed	59.93	62.50	62.67	64.78	45.00	57.00
	(16.87)	(15.36)	(22.54)	(10.59)	(20.43)	(7.07)
17. Memory for Words	73.20	79.13	75.67	72.67	62.40	71.50
	(11.01)	(8.51)	(4.76)	(10.25)	(16.46)	(7.78)
18. Rapid Picture Naming	67.60	66.50	71.67	72.11	58.80	61.50
	(15.40)	(16.13)	(17.74)	(17.10)	(9.36)	(6.36)

Note. F= Functional readers; L = Lexical readers; NF = Nonfunctional readers; ND = Nondecoders; NR= Nonreaders. Standard scores with mean = 100 and standard deviation = 15. [*]For the Visual-Auditory Delayed subtest, the Nondecoders and Nonreaders did not attempt this due to the number of errors they made on the initial learning subtest (subtest 2).

[a]Difference between subgroups approached significance [$F(4,25) = 3.638$, $p = .018$]

[b]Difference between subgroups approached significance [$F(4, 25) = 3.423$, $p = .023$]

[c]Significant difference between subgroups [$F(4,25) = 5.586$, $p < .005$]

[d]Significant difference between subgroups [$F(4,25) = 5.337$, $p < .005$]

In summary, by analysing reading performance on the subscales of irregular words and nonwords, different patterns of reading performance were identified. These included Functional readers (average, for end of 5th Grade, performance on both word types), Lexical readers (average performance on irregular words but impaired on nonwords) and a Mixed group (impaired performance on both word types). The Mixed group was further categorised into Nonfunctional readers (reading below a functional level for both word types), Nondecoders (can read a low level of irregular words but cannot decode nonwords) and two readers were classified as Nonreaders. The difference between the subgroups was not accounted for by different IQ levels of the subgroups. The subgroups differed on cognitive measures of phonemic awareness and working memory and there was evidence to suggest that verbal comprehension and visual-auditory learning skills also varied for the subgroups.

Discussion

This research aimed to investigate regular, irregular and nonword reading ability in WS to determine whether individuals with WS could attain a functional level of literacy, assessed as being equivalent to the end of Year 5 primary school level, and to examine the relationship between reading ability with adaptive and intellectual functioning. In addition to intellectual functioning, cognitive domains known to be associated with reading in typical readers was assessed to determine whether the cognitive correlates of reading ability in WS are similar to that of typically developing individuals. In addition, patterns of reading ability were assessed individually using Dual Route theory to determine whether there was evidence for particular strengths and/or weaknesses in lexical versus nonlexical reading and whether individuals with WS showed a universal reading profile. The results are discussed in turn, closely following the aims outlined at the end of the introduction.

Functional reading in WS

Out of 30 individuals with WS, approximately one third (27%) read at a level of functional literacy across both irregular and nonwords and a further 20% read irregular words at a functional level. Functional was defined as reading within one standard deviation of the mean or end of Year 5 primary school students. Note that this does not say anything about their ability to read at the level of their chronological age. Of the 28 individuals who were able to read, the average approximate reading age for regular words was 8 years 3 months (range was from 6 years 2 months to over 11 years 5 months), which is well below the average chronological age of 21 years. This is consistent with previous studies which have reported the mean reading age in WS to be around the 7 to 9 year age level and typically below the level of their chronological age (Howlin et al., 1998; Menghini et al., 2004; Temple, 2006; Udwin et al., 1987, Udwin et al., 1996). This reading level, on average, is more similar to (but slightly above) the mental age level of the group.

Higher reading skills are associated with greater levels of everyday independence, particularly in the areas of Written communication and Community living

This paper is the first to report on the relationship between reading ability and adaptive functioning in WS. Higher reading ability on face-to-face testing was found to be associated with higher outcomes on the Expressive and Written subdomains of the Vineland II. It was not surprising to find the Written subdomain to be related to all reading subscales on the CC2 and supporting our hypothesis, as this subdomain aims to assess alphabetic knowledge and typical reading and writing abilities in daily life, such as ability to read a newspaper. Higher Expressive communication skills, such as how an individual uses words and sentences to gather and provide information (Sparrow et al., 2005), was also associated with higher reading abilities on face-to-face testing across all reading subscales on the CC2. This is consistent with previous studies in typically developing individuals, which have shown that language processing skills are related to reading (see Mann, 2003). Perhaps more importantly, higher reading abilities were also found to be associated with higher outcomes on the Community subdomain (within the Daily Living Skills domain). This subdomain measures how an individual operates within their community and includes everyday tasks such as understanding and following rules, the use of time and dates, money, telephone, computers, and job skills. This relationship appears to be independent of intellectual functioning, as the relationship between reading and Community living remained significant when FSIQ was statistically controlled. This suggests that individuals with WS who have a functional level of literacy are also found to be functioning more independently on everyday tasks. It is clear that being able to read would help an individual to be able to undertake a range of daily tasks, such as being able to choose an item from a menu, identify products when shopping, read signs and timetables to enable independent travel, or use technology such as a computer. Job skills are also assessed under this subdomain, so better reading skills may also result in more vocational opportunities.

We are only aware of one other study (Howlin et al., 1998) that has included a measure of adaptive functioning on the VABS and reading abilities in WS. The authors noted that adaptive skills were low; however, they did not statistically compare the adaptive and reading abilities, perhaps due to psychometric limitations of the VABS only providing age equivalent scores at the subdomain level. Due to the lack of attainment in academic skills, Howlin et al. (1998) concluded that to improve academic outcomes, a greater focus should be on their academic problems and adapting the education curriculum accordingly. Our findings suggest that a focus on developing reading skills would also be advantageous and may generalise to higher independence in daily life.

The significant correlations between adaptive functioning and reading in our study may indicate that focussing resources on improving reading might have positive effects on adaptive functioning for individuals with WS. However, actual training studies are required to investigate if this is indeed true. Udwin et al. (1996) completed a longitudinal study (from Udwin et al., 1987) and reported a small non-significant increase of around 11 months in reading ability over eight and a half years. The authors suggested that the negligible increase may be due to limited education opportunities available to individuals after they left formal schooling or, alternatively, this may suggest a ceiling of reading ability for WS individuals at around the 7 to 9 year level. They recommended that older adolescents with WS should perhaps focus on developing functional daily living skills (e.g., independent travel or handing money) rather than academic skills (however, the authors also noted that it was difficult to make direct comparisons as different reading tests were used on each occasion, the Neale Analysis of Reading Ability and the WORD basic reading subtest at follow-up). However, it is possible that the rate of reading attainment and development may be much slower in WS. As has been noted for individuals with Down syndrome and intellectual disability more generally, when students move into the secondary school level, the focus of the curriculum tends to move to more practical life skills, however, it may be argued that they would continue to benefit from rigorous reading instruction throughout high school and beyond to reach a level of functional literacy to enable greater vocational, daily living and recreational opportunities (Bochner et al., 2001).

Correlates of Reading Ability and Intellectual and Cognitive Ability for the Group

Using an absolute difference method (Siegal, 1992), the majority of individuals (from 61-89% depending on reading subscale) showed no significant difference between their GIA (measure of overall intellectual functioning) and reading abilities across the three CC2 reading subscales. There were a smaller number of individuals whose level of intellectual functioning was significantly lower than their reading ability, but no individual showed a higher level of intellectual functioning compared to their reading level. Therefore, individuals in our study attained a level of reading either above or commensurate with their level of intellectual functioning. This finding indicates that learning to read should be an aim for all individuals regardless of their IQ level.

Regarding cognitive abilities, an advantage of using the WJ III COG is that, as all the subtests included in the study were taken from a single standardised battery, all of the subtests share a common metric and the same normative reference group, which allows more valid comparison of abilities (Mervis & Robinson, 1999). For the group, the three largest associations for both lexical and nonlexical reading were for visual-auditory (associative) learning, phonemic awareness (sound blending), and working memory (numbers reversed). Working memory made a significant and unique contribution to nonlexical reading, but not for lexical reading. Verbal comprehension was also associated with all three reading subscales. These results are in line with studies which have also found associations in reading in WS with phonological awareness (e.g., Laing et al., 2001; Levy & Antebi, 2004; Levy et al., 2003; Menghini et al., 2004). Previous WS reading studies have included tasks assessing the ability to refresh phonological stores using digit span tasks or word span tasks (e.g., Laing et al., 2001; Levy & Antebi, 2004). However, studies have not included more demanding working memory tasks, requiring simultaneous processing and manipulation or reorganising verbal information. Our results indicate that working memory, perhaps more so than phonological stores is highly predictive of reading abilities in WS. It will therefore be important to consider working memory limitations when implementing reading instruction for WS. Interventions targeting working memory may also generalise to greater improvements in reading.

In contrast to typically developing children, rapid automatic naming (RAN) was not associated with reading ability in our study. Other studies have also failed to find this association in WS (Dessalegn et al., 2012; Levy et al., 2003; Levy & Antebi, 2004), while Laing did find an association with RAN (Laing et al., 2001). The different results across studies may be due to the diverse items participants are requested to name, or to heterogeneity between samples, as has been shown in studies of cognitive abilities in WS (Jarrold et al., 1998; Pezzini et al., 1999; Porter & Coltheart, 2005; Tassabehji et al., 1999). In contrast to other RAN tasks, the WJ III COG uses a set of unique items rather than have a small set of items, repeated. As this is a speeded test, this may have placed increased demands on retrieval abilities for WS participants. A nonverbal subtest, Analysis Synthesis (part of the Fluid Reasoning cluster) correlated with lexical and nonlexical reading and the speeded Visual Matching subtest was associated with nonlexical (nonword reading). This was an unexpected finding as visual abilities have not been found to be related to reading in typically developing individuals (Evans et al., 2001). However, some studies have reported an association between nonverbal abilities and reading in WS (Dessalegn et al., 2012; Laing et al., 2001; Levy et al., 2003) which indicates that visual-spatial weaknesses in WS may contribute to their reading difficulties. The Visual Matching subtest requires individuals to quickly scan through a row and identify matching numbers. This task places demands on visual scanning ability, which is difficult in WS. Further investigation of visually related difficulties is an area in need of further investigation as it would have implications for reading instruction.

As the same underlying cognitive skills which have been found to be related to reading in typically developing children have also been found to be associated with reading in WS, this implies that individuals with WS are likely to benefit from similar teaching strategies. However, the specific cognitive limitations known to be a weakness in WS will need to be taken into account and teaching strategies modified accordingly. For example, to reduce the burden on working memory abilities, the use of picture cues would be recommended (see Channell, Loveall, & Conners, 2013) and for visual-spatial difficulties the use of highlighting discriminating features in visual stimuli (see Riby & Porter, 2010).

Reading patterns in WS

It is important to determine whether the reading difficulties observed in individuals with WS are universal, for if this were the case, then a "one-size-fits-all" approach to reading instruction may be appropriate. However, in this study we confirmed the heterogeneity of reading profiles in WS by using a subtyping procedure based on the Dual Route theory of reading. Subtyping of reading difficulties has been used widely in the developmental dyslexia population of typically developing children and the CC2 test was designed specifically for this purpose (Castles et al., 2009; Edwards & Hogben, 1999). Our sample included non-readers who could not read any words at all, consistent with previous WS studies (Howlin et al., 1998; Laing et al., 2001; Levy & Antebi, 2004; Levy et al., 2003; Udwin et al., 1987) and individuals who could not decode any nonwords, which has also been reported previously (Becerra et al., 2008 as cited in Mervis, 2009; Laing et al, 2001; Levy & Antebi, 2004; Levy et al., 2003). Subgroups of individuals who could read irregular and nonwords, but at different ability levels (either at or below the functional level of literacy or end of Year 5 primary school) were identified.

Interestingly, there were no individuals in our sample with relative strengths in nonword reading compared to irregular word reading. This is at odds with Temple (2006) who reported a group of eight children with WS whose reading age was at the level of their mental age (MA from 5;7 to 7;9) and no impairment in nonword reading relative to mental age matched controls; however, irregular and high frequency word reading was impaired. It is unclear why our sample did not contain any individuals with a relative strength in nonlexical reading. In fact, our findings indicate that if only one reading scale is impaired, it tends to be the nonword reading subscale. It is possible that early instructional methods for these participants may have focused on sight word reading (perhaps having been exposed to a whole-language approach to teaching reading) and have, therefore, not received explicit, systematic phonics instruction. Background information about the type of reading instruction for each participant would be useful here, both in the current study and in Temple (2006). It is certainly possible that Temple's UK sample received different educational methods of reading instruction to the current Australian population. Temple (2006) hypothesised that this pattern may either reflect the resolution of deep dyslexia with the acquisition of phonological skills leading to an over-reliance on nonlexical reading, or may reflect the fact that there are distinct subtypes of reading disorders in WS. Our findings would appear to confirm the latter.

Our findings highlight the need to analyse reading ability at the individual level, as well as the group level. While group trends contribute to knowledge about the typical level of reading ability attained in WS, at the group level, important information is lost, as it is unknown whether the readers who perform better on one scale (e.g., nonword reading) are the same individuals who perform higher on word reading. Previous case studies have taken the approach of analysing individual performance (Barca et al., 2009; Dessalegn et al., 2012; Temple, 2003) and two group studies provided individual scores as well as group means (Levy & Antebi, 2004; Pagan et al., 1987). Further, two group studies have considered reading subgroups based on IQ score ranges (Levy & Antebi, 2004; Levy et al., 2003). While Levy and Antebi (2004) looked at reading of words compared to nonwords, their main focus was on decoding and the ability to read nonwords. This is, thus, the first group study in WS which has systematically analysed each individual's reading performance to enable profiling of relative strengths and weaknesses in lexical and nonlexical performance. Overall, given the heterogeneity of the sample, it does not seem prudent to prescribe one intervention for all individuals with WS.

Cognitive correlates of lexical and nonlexical reading in WS

Levy et al. (2003, see also Levy, 2011) proposed that IQ is the strongest predictor of word decoding ability in WS, however, our results indicate that IQ alone cannot explain the difference between lexical and nonlexical reading abilities within the different reading subgroups. This finding is consistent with research which has reported that FSIQ is not a useful predictor of reading ability in typically developing children (Siegel, 1992; McArthur et al., 2013) nor in children with intellectual disability (Conners et al., 2001). Levy et al. (2003, see also Levy, 2011) noted that the underlying cognitive processes of reading, such as phonological awareness, rely on meta-cognitive procedures which, in turn, relate to level of intellectual functioning. The utility of a FSIQ score in WS is somewhat limited due to the variability in their cognitive functioning and a single IQ score may mask individual variability. As such, we thought it would perhaps be more meaningful to consider individual cognitive skills which have been shown to relate to reading in typically developing children and also consider the possible impact of areas of specific weakness in WS, such as visual-spatial or working memory (WM) deficits. For example, Conners et al., (2001) reported that for children with unspecified intellectual disability, being able to rehearse phonological codes in WM was more important than FSIQ, language ability or phonemic awareness in success in learning to read. Our study found that the cognitive skills which were found to be significantly different between the reading subgroups were the tasks of sound blending (Phonemic Awareness measure) and numbers reversed (Working Memory measure). In addition, Verbal Comprehension and Visual-Auditory Learning came close to significantly differentiating between the reading subgroups. Therefore, the same skills which were found to relate to reading for the group were also found to differentiate between the reading subgroups. The Functional Readers who could read via both lexical and nonlexical routes had the highest mean score on all of these cognitive abilities. This highlights again that similar cognitive skills required for better reading abilities in WS are also found in typical readers. The main clinical implications here are that reading instruction for typically developing readers may be as successful for individuals with WS, however, the particular cognitive strengths and weaknesses identified in individuals with WS also need to be taken into account and modifications and adjustments made to teaching instruction accordingly.

Limitations and future directions

Although this was not the aim of the study, a limitation of using the CC2 test was that we were not able to provide information for the majority of participants, as to whether they could read at the level expected for their chronological age (although three participants' ages fell within the normative age range and for all three, reading ability was below the level of chronological age but higher than mental age). Previous studies have utilised a variety of standardised and experimental word lists. These lists can vary on numerous factors, such as the inclusion of irregular and/or regular words and the length and frequency of the words, all of which can result in variations in the rate at which the test increases in difficulty. The CC2 test was designed to minimise some of these potentially confounding factors. The study also did not assess reading comprehension, and some studies have shown this to be an area of weakness in WS (e.g., Laing et al., 2001; Menghini et al., 2004). Further research is needed in this important area, to ascertain whether individuals who can read at a functional level are able to understand the information they are reading (along with other aspects of performance such as reading fluency). Therefore, along with incorporating measures from adaptive functioning scales, more ecologically valid tests may need to be designed. For example, Bochner et al. (2001) assessed functional literacy skills in young adults with Down syndrome through informal measures including recognition of everyday signs (e.g., exit, stop) and identifying the time and day of a program from a television guide.

A case study by Dessalegn et al. (2012) investigated visual-spatial deficits on reading ability for two individuals with WS and found difficulties with visual orientation were related to an increase in letter orientation and letter ordering errors for one individual. While a systematic analysis of errors was not undertaken in this study as the CC2 word list was not specifically designed to capture evidence of visual sequencing or orientation errors, anecdotally, there was some evidence of visual errors made by some participants. For example, nerve →never; borp →brop. Therefore, further investigation of visual type errors in WS is warranted. In support of possible underlying spatial/visual contribution to reading, we did find positive, medium correlations between reading and WJ III COG Test 6, Visual Matching and Test 3, Spatial Relations which approached significance (the large correlation between nonword reading and Visual Matching was significant).

Conclusion

In conclusion, this is the first study to demonstrate a relationship between functional reading ability and everyday living skills for individuals with WS. The study found that around one third of the group of WS individuals demonstrated a functional level of literacy on basic word reading. There was however, evidence of considerable heterogeneity in abilities and analysing individual performance revealed subtypes of reading ability across the lexical and nonlexical reading routes. Where there was a difference between abilities, the nonlexical route showed greater impairment compared to the lexical route. Intellectual functioning (IQ) was not found to account for the differences between the subgroups, indicating that all individuals with WS should be provided with appropriate reading instruction regardless of IQ. The clinical implication of this is that appropriate early intervention for literacy attainment is important to ensure individuals with WS have the opportunity to develop their skills to as high a level as possible and in doing so, these abilities may well generalise to other life skills and independence in daily living. A complete neuropsychological assessment would be recommended to assess cognitive strengths and weaknesses, particularly for areas which are related to reading (e.g., associative learning, phonemic awareness, working memory) and for cognitive skills known to be a relative weakness in WS (e.g., visual-spatial skills) to tailor the literacy instruction for the individual accordingly. It is also important to remember that appropriate reading instruction requires *both* phonics training combined with sight word training. For typically developing readers it is recommended that phonics instruction be both *explicit* (where letter-sound relationships are taught directly) and *systematic* (with a defined scope and sequence of letter-sound instruction) as opposed to letter-sound relationships being taught incidentally while engaging with text (an embedded or literature-based approach). This may be even more important for individuals with WS who appear to have greatest difficulty with decoding skills and they may require additional instruction and practice to ensure that letter-sound knowledge can be internalised. By improving literacy outcomes in individuals with WS, the aim is to improve their everyday living skills and as has been demonstrated in the significant relationship between reading skills and adaptive functioning in our study. Most encouragingly, there is evidence to suggest that this may generalise to improve functioning in many aspects of an individual's everyday life.

References

Arnell, K.M., Klein, R.M., Joanisse, M.F., Busseri, M.A., & Tannock, R. (2009). Decomposing the relation between rapid automatized naming (RAN) and reading ability. *Canadian Journal of Experimental Psychology, 63,* 173-184.

Australian Bureau of Statistics. (2006). *Census of Population and Housing: Index of Relative Socio-Economic Disadvantage (IRSD).* Retrieved from http://www.abs.gov.au/AUSSTATS/abs@.nsf/DetailsPage/2033.0.55.0012006?OpenDocument.

Barca, L., Bello, A., Volterra, V., & Burani, C. (2009). Lexical-semantic reading in shallow orthography: Evidence from a girl with Williams syndrome. *Reading Writing, 23,* 569-588.

Bochner, S., Outhred, L., & Pieterse, M. (2001). A study of functional literacy skills in young adults with Down syndrome. *International Journal of Disability, Development and Education, 48,* 67-90.

Brock, J. (2007). Language abilities in Williams syndrome: A critical review. *Development and Psychopathology, 19,* 97-127.

Castles, A., & Coltheart, M. (2004). Is there a causal link from phonological awareness to success in learning to read? *Cognition, 91,* 77-111.

Castles, A., Coltheart, M., Larsen, L., Jones, P., Saunders, S., & McArthur, G. (2009). Assessing the basic components of reading: A revision of the Castles and Coltheart test with new norms. *Australian Journal of Learning Difficulties, 14,* 67-88.

Channell, M.M., Loveall, S.J., & Conners, F.A. (2013). Strengths and weaknesses in reading skills of youth with intellectual disabilities. *Research in Developmental Disabilities, 34,* 776-787.

Colenbrander, D., Nickels, L., & Kohnen, S. (2011). Nonword reading tests: A review of the available resources. *Australasian Journal of Special Education, 35,* 137-172.

Coltheart, M., Rastle, K., Perry, C., Langdon, R., & Ziegler, J. (2001). DRC: A dual route cascaded model of visual word recognition and reading aloud. *Psychological Review, 108,* 204-256.

Conners, F.A., Atwell, J.A., Rosenquist, C.J., Sligh, A.C. (2001). Abilities underlying decoding differences in children with intellectual disability. *Journal of Intellectual Disability Research, 45,* 292-299.

Conners, F.A., Moore, M.S., Loveall, S.J., Merrill, E.C. (2011). Memory profiles of Down, Williams, and Fragile X syndromes: Implications for reading development. *Journal of Developmental & Behavioral Pediatrics, 32,* 405-417.

Conners, F.A., Rosenquist, C.J., Sligh, A.C., Atwell, J.A., & Kiser, T. (2006). Phonological reading skills acquisition by children with mental retardation. *Research in Developmental Disabilities, 27*, 121-137.

Cupples, L., & Iacono, T. (2000). Phonological awareness and oral reading skill in children with Down syndrome. *Journal of Speech, Language, and Hearing Research, 43,* 595-608.

Cupples, L., & Iacono, T. (2002). The efficiency of “whole word” versus “analytic” reading instruction for children with Down syndrome. *Reading and Writing: An Interdisciplinary Journal, 15,* 549-574.

Dessalegn, B., Landau, B., & Rapp, B. (2012). Consequences of severe visual-spatial deficits for reading acquisition: Evidence from Williams syndrome. *Neurocase: The Neural Basis of Cognition, iFirst,* 1-20.

Edwards, V.T., & Hogben, J.H. (1999). New norms for comparing children’s lexical and nonlexical reading: A further look at subtyping dyslexia. *Australian Journal of Psychology, 51,* 37-49.

Evans, J.J., Floyd, R.G., McGrew, K.S., & Leforgee, M.H. (2001). The relations between measures of Cattell-Horn-Carroll (CHC) cognitive abilities and reading achievement during childhood and adolescence. *School Psychology Review, 31,* 246-262.

Ewart, A.K., Morris, C.A., Atkinson, D., Jin, W., Sternes, K., Spallone P., Stock, A.D., Leppert, M., & Keating, M.T. (1993). Hemizygosity at the elastin locus in a developmental disorder, Williams syndrome. *Nature Genetics, 5,* 11–16.

Gombert, J. (2002). Children with Down syndrome use phonological knowledge in reading. *Reading and Writing: An Interdisciplinary Journal, 15,* 455-469.

Heinze, E.G., & Vega, F.C. (2008). Aprendizaje de la lectura en los ninos con syndrome de Williams [Abstract]. *Psicothema, 20,* 672-677.

Heller, R., Rauch, A., Lüttgen, S., Schröder, B. & Winterpacht, A. (2003). Partial deletion of the critical 1.5Mb interval in Williams-Beuren syndrome. *Journal of Medical Genetics, 40,* e99.

Howlin, P., Davies, M., & Udwin, O. (1998). Cognitive functioning in adults with Williams syndrome. *Journal Child Psychology & Psychiatry, 39,* 183-189.

Jarrold, C., Baddeley, A.D., & Hewes, A.K. (1998). Verbal and non-verbal abilities in the Williams syndrome phenotype: Evidence for diverging developmental trajectories. *Journal of Child Psychology and Psychiatry, 39,* 511-523.

Karmiloff-Smith, A., Broadbent, H., Farran, E.K., Longhi, E., D'Souza, D., Metcalfe, K., Tassabehji, M., Wu, R., Senju, A., Happe, F., Turnpenny, P., & Sansbury, F. (2012). Social cognition in Williams syndrome: Genotype/phenotype insights from partial deletion patients. *Frontiers in Psychology, 3,* 1-8.

Laing, E. (2002). Investigating reading development in atypical populations: The case of Williams syndrome. *Reading and Writing: An Interdisciplinary Journal, 15,* 575-587.

Laing, E., Hulme, C., Grant, J., & Karmiloff-Smith, A. (2001). Learning to read in Williams syndrome: Looking beneath the surface of atypical reading development. *Journal of Child Psychology and Psychiatry, 42*, 729-739.

Levy, Y. (2011). IQ predicts word decoding skills in populations with intellectual disabilities, *Research in Developmental Disabilities, 32,* 2267-2277.

Levy, Y., & Antebi, V. (2004). Word reading and reading-related skills in Hebrew-speaking adolescents with Williams syndrome. *Neurocase, 10,* 444-451.

Levy, Y., Smith, J., & Tager-Flusberg, H. (2003). Word reading and reading-related skills in adolescents with Williams syndrome. *Journal of Child Psychology and Psychiatry, 44*, 576-587.

Lowery, M.C., Morris, C.A., Ewart, A., Brothman, L.J., Zhu, X.L., Leonard, C.O., Carey, J.C., Keating, M., & Brothman, A.R. (1995). Strong correlation of elastin deletions, detected by FISH with Williams syndrome: Evaluation of 235 patients. *American Journal of Human Genetics, 57,* 49-53.

Mann, V.A. (2003). Language processes: Keys to reading disability. In H.L. Swanson, K.R. Harris & S. Graham (Eds), *Handbook of learning disabilities* (pp. 213-228).New York, NY: Guilford Press.

Martens, M.A., Wilson, S.J., & Reutens, D.C. (2008). Research review: Williams syndrome: A critical review of the cognitive, behavioral, and neuroanatomical phenotype. *The Journal of Child Psychology and Psychiatry, 49,* 576-608.

Mather, N. & Woodcock, R.W. (2001). Examiner's Manual. *Woodcock-Johnson III Tests of Cognitive Abilities.* Rolling Meadows, IL: Riverside Publishing.

McArthur, G., Kohnen, S., Larsen, L., Jones, K., Anandakumar, T., Banales, E., & Castles, A. (2013). Getting to grips with the heterogeneity of developmental dyslexia. *Cognitive Neuropsychology, 30,* 1-24.

McGrew, K., & Woodcock, R. W. (2001). Technical Manual. *Woodcock-Johnson III.* Rolling Meadows, IL: Riverside Publishing.

Menghini, D., Verucci, L., Vicari, S. (2004). Reading and phonological awareness in Williams syndrome. *Neuropsychology, 18,* 29-37.

Mervis, C.B. (2009). Language and literacy development of children with Williams syndrome. *Topics in Language Disorders*, *29*, 149-169.

Mervis, C.B., & John, A.E. (2010). Cognitive and behavioral characteristics of children with Williams syndrome. *American Journal of Medical Genetics Part C (Seminars in Medical Genetics), 154C*, 229-248.

Mervis, C.B., & Klein-Tasman, B.P. (2004). Methodological issues in group-matching designs: Alpha levels for control variable comparisons and measurement characteristics of control and target variables. *Journal of Autism and Developmental Disorders, 34,* 7-17.

Mervis, C. B., & Robinson, B. F. (1999). Methodological issues in cross-syndrome comparisons: Matching procedures, sensitivity (*Se*), and specificity (*Sp*). Commentary on M. Sigman & E.Ruskin, Continuity and change in the social competence of children with autism, Down syndrome, and developmental delays. *Monographs of the Society for Research in Child Development, 64,* 115–130.

Moore, D.M., Porter, M.A., Kohnen, S., & Castles, A. (2012). Detecting different types of reading difficulties: A comparison of tests. *Australasian Journal of Special Education, 36,* 112-133.

Nilsen, E., & Bourassa, D. (2008). Word learning performance in beginning readers. *Canadian Journal of Experimental Psychology, 62,* 110-116.

Pagan, R.A., Bennett, F.C., LaVeck, B., Stewart, K.B., & Johnson, J. (1987). Williams syndrome: Features in late childhood and adolescence. *Pediatrics, 80,* 85-91.

Pezzini, G., Vicari, S., Voltera, V., Milani, L., & Ossella, M.T. (1999). Children with Williams syndrome: Is there a single neuropsychological profile? *Neuropsychologia, 15,* 141-155.

Porter, M.A. & Coltheart, M., (2005). Cognitive heterogeneity in Williams syndrome. *Developmental Neuropsychology, 27,* 275-306.

Porter, M.A., Dobson-Stone, C., Kwok, J.B.J., Schofield, P.R., Beckett, W., & Tassabehji, M. (2012). A role for transcription factor GTF2IRD2 in executive function in Williams-Beuren syndrome. *PLoS ONE, 7,* e47457. DOI: 10.1371/journal.pone.0047457.

Riby, D.M., & Porter, M.A. (2010). Chapter 5 – Williams syndrome. *Advances in Child Development and Behavior, 39,* 163-209.

Schrank, F.A., Flanagan, D.P., Woodcock, R.W., & Mascolo, J.T. (2002). *Essentials of WJ III Cognitive Abilities Assessment.* New York: John Wiley & Sons

Schrank, F. A., & Woodcock, R. W. (2009). WJ III Normative Update Compuscore and Profiles Program, *Australian Adaptation;* [WJ III NU (AA)], [Computer software]. Adaptation of the *Woodcock-Johnson III Normative Update* (WJ III NU) Compuscore and Profiles Program. Rolling Meadows, IL: Riverside Publishing.

Schubert, C. (2009). Review: The genomic basis of Williams-Beuren syndrome. *Cellular and Molecular Life Sciences, 66,* 1178-1197.

Siegel, L.S. (1992). An evaluation of the discrepancy definition of dyslexia. *Journal of Learning Disabilities, 25,* 618-629.

Sparrow, S.S., Balla, D., & Cicchetti, D. (1984) *Vineland Adaptive Behavior Scales.* Circle Pines, MN: American Guidance Service.

Sparrow, S.S., Cicchetti, D.V., & Balla, D.A. (2005). *Vineland Adaptive Behavior Scales* (2nd Ed.). Survey Forms Manual. AGS Publishing, Circle Pines, MN: AGS Publishing.

Strattman, K., & Hodson, B.W. (2005). Variables that influence decoding and spelling in beginning readers. *Child Language Teaching and Therapy, 21,* 165-190.

Strømme, P., Bjørnstad, P.G., Ramstad, K. (2002). Prevalence estimation of Williams syndrome. *Journal of Child Neurology, 17,* 269-271.

Stuart, M., Masterson, J., & Dixon, M. (2000). Spongelike acquisition of sight vocabulary in beginning readers? *Journal of Research in Reading, 23,* 12-27.

Tassabehji, M., Metcalfe, K., Karmiloff-Smith, A., Carette, M.J., Grant, J., Dennis, N., et al. (1999). Williams syndrome: Use of chromosomal microdeletions as a tool to dissect cognitive and physical phenotypes. *American Journal of Human Genetics, 64,* 118-125.

Tassabehji, M. (2003). Williams-Beuren syndrome: A challenge for genotype-phenotype correlations. *Human Molecular Genetics, 12,* r229-r237.

Temple, C. (2003). Deep dyslexia in Williams syndrome. *Journal of Neurolinguistics, 16,* 457-488.

Temple, C. (2006). Developmental and acquired dyslexias. *Cortex, 42,* 898-910.

Udwin, O., Davies, M., Howlin, P. (1996). A longitudinal study of cognitive abilities and educational attainment in Williams syndrome. *Developmental Medicine and Child Neurology, 38,* 1020-1029.

Udwin, O. & Yule, W. (1991). A cognitive and behavioural phenotype in Williams syndrome. *Journal of Clinical and Experimental Neuropsychology, 13,* 232-244.

Udwin, O., Yule, W., & Martin, N. (1987). Cognitive abilities and behavioural characteristics of children with idiopathic infantile hypercalcaemia. *Journal of Child Psychology and Psychiatry, 28,* 297-309.

Vineland II ASSIST Scoring and Reporting System for Survey Forms [Computer software]. (2008). San Antonio, TX: Pearson.

Verucci, L., Menghini, D., Vicari, S. (2006). Reading skills and phonological awareness acquisition in Down syndrome. *Journal of Intellectual Disability Research, 50,* 477-491.

Wagner, R.K., & Torgesen, J.K. (1987). The nature of phonological processing and its causal role in the acquisition of reading skills. *Psychological Bulletin, 101,* 192-212.

Wheldall, K., & Watkins, R. (2004). Literacy levels of male juvenile justice detainees. *Educational Review, 56,* 3-11.

Windfuhr, K.L., & Snowling, M.J. (2001). The relationship between paired associate learning and phonological skills in normally developing readers. *Journal of Experimental Child Psychology, 80,* 160-173.

Wise, J.C., Sevcik, R.A., Romski, M., & Morris, R.D. (2010). The relationship between phonological processing skills and word and nonword identification performance in children with mild intellectual disabilities, *Research in Developmental Disabilities, 31,* 1170-1175.

Woodcock, R.W., McGrew, K.S., & Mather, N. (2001). *Woodcock-Johnson III Tests of Cognitive Abilities.* Rolling Meadows, IL: Riverside Publishing.

Thesis Conclusion

This thesis explored adaptive functioning in individuals with WS. The overall aims of the thesis were: 1) to review the literature on adaptive functioning in WS; 2) to profile adaptive functioning skills of individuals with WS and investigate the relationship between adaptive functioning and demographic and environmental factors, including gender, chronological age, intellectual and cognitive functioning and the family environment, and 3) to investigate lexical and nonlexical reading abilities in WS and to consider the relation between reading and adaptive functioning. In line with these aims, the first chapter was a systematic review of the literature regarding adaptive functioning in WS, the second chapter was an empirical study of adaptive functioning skills in WS and the third chapter was an empirical study of functional literacy skills in WS. Overall, findings from Chapter 1 indicated that further information is needed regarding adaptive functioning skills across the life-span for individuals with WS and to consider environmental factors which may influence development of adaptive abilities. Such research was considered important, as the level of support that an individual with WS requires to develop independence may increase as they get older, rather than decrease, as would be expected for typically developing individuals.

Findings from the empirical study of Chapter 2 provided evidence of heterogeneity in adaptive skills for individuals with WS, with different patterns of relative strengths and weaknesses observed, along with differences between younger individuals still at school, compared to older individuals who had completed formal schooling. This suggests that formal schooling may provide a higher level of support and a structured environment which may be beneficial in the development of adaptive skills. Furthermore, this empirical study is the first to show that some aspects of the family environment, in particular the family's participation in social, recreational and cultural/ intellectual activities and pastimes, can be related to higher adaptive skills, perhaps by providing individuals with important learning experiences and opportunities. The findings also suggest that higher levels of structure and routine within the family environment can result in higher adaptive abilities. Perhaps this style of household replicates the school environment which naturally provides rules, routines and set procedures. Furthermore, future research should investigate the relationship between executive dysfunction and adaptive functioning skills. Executive functioning is thought to be impaired in individuals with WS. Executive functioning requires higher order cognitive functions such as impulse control, inhibition, working memory, attention, flexibility and planning; skills which are required to successfully cope with new and difficult situations. Executive dysfunction may relate to some of the difficulties with attaining functional independence and some of the problem behaviours evident in WS. For example, difficulties adapting and coping with changes in routine, inhibiting asking embarrassing questions in public, or being able to manage and use money in a bank account responsibly.

The empirical study of Chapter 3 revealed heterogeneity in reading abilities in WS with different patterns of performance evident across lexical and nonlexical reading. The findings indicate that reading instruction should be provided for all individuals with WS regardless of their intellectual functioning, as reading ability was not related to IQ. As noted for adaptive functioning skills, the time spent in formal schooling may provide a higher level of support and a structured environment which may be beneficial in the development of adaptive skills Also; higher levels of reading ability were found to relate to higher outcomes in functioning within the community. Therefore, reading is an important skill to develop for individuals with WS, as it may generalise to improvements in independence across many everyday living skills. The cognitive skills which were found to be predictors of reading in WS are also known to be associated with reading in typically developing individuals, indicating that reading instruction recommended for typical readers, incorporating both decoding and visual recognition, would seem to be suitable for individuals with WS.

Findings from both Chapter 2 and Chapter 3 highlight the need for ongoing support for adults with WS, especially once they no long have the routine and structure provided by the formal school setting. There is a need for individuals to be challenged with appropriate work and recreational activities to enable them to continue to strive to live as independently as possible. Ongoing educational, social, vocational and vacational opportunities, such as transition to work programs, suitable TAFE training courses, and recreational groups are indicated to ensure they continue to have the opportunity for socialising and further development of their academic, adaptive and social skills. This is also important for the opportunity for ongoing practice in literacy skills. Lack of exposure to further education and employment may lead to reduced opportunities and therefore a plateauing in literacy abilities with increasing age for individual with WS.

The research highlights that group level performance does not necessarily reflect the heterogeneity which is evident in WS in areas of adaptive functioning and reading abilities. Therefore, for adaptive functioning, there is a need to go beyond an overall score which may mask information about relative strengths and weaknesses, and to investigate domain and subdomain performance and consider individual variability. In regards to reading abilities, analysis of performance on lexical and nonlexical reading is recommended. The clinical implication here includes the need for comprehensive neuropsychological assessment of intellectual, cognitive, adaptive functioning and literacy skills in individuals with WS to be able to provide relevant information about strengths and weaknesses and subsequent support needs. While appropriate support may be available while an individual is within a structured learning environment, such as the school setting, individuals with WS may require formal, structured support and assessment beyond the school years to enable them to continue to develop their daily living skills and improve their literacy outcomes.

Knowledge of the cognitive and behavioural phenotype of individuals with WS need to be integrated with results of adaptive functioning, as cognitive and behavioural difficulties have the potential to impact on adaptive skills, while relative strengths can be integrated into learning and intervention programs. For example, pragmatic language difficulties can affect social communication and working memory problems and can result in difficulties completing multi-step instructions. Interventions here may include social skills or input from a speech and language therapist to further develop social language skills, and breaking down tasks into smaller steps and providing appropriate reminders (e.g., visual prompts). Higher levels of behaviour problems, which may be due to executive dysfunction, can result in impulsivity or perseveration, leading to problems with conversation skills, such as interrupting others, repeating or getting ‘stuck’ on particular topics of self-interest. Therefore, clinicians should not make assumptions about functioning despite an individual with WS presenting as outgoing and friendly, as they may need explicit assistance in social skills development and organised recreational and social events for older individuals. Detailed assessment of adaptive functioning is also required for diagnostic purposes, as an IQ score will not provide the necessary information to determine an individual’s support needs. This thesis showed that behaviour difficulties can relate to adaptive functioning and interventions here may include behaviour programs and working with families to help overcome behaviour difficulties. Through more targeted interventions in the areas of behaviour and reading, improvements may generalise to greater independence in life skills.

Overall, the main contributions of this thesis were in demonstrating that adaptive functioning and literacy skills are heterogeneous in WS and individuals are likely to require additional support to help them continue to develop their skills, even after they have completed their formal years of schooling. Future research should now focus on efficacy of teaching strategies for individuals with WS to determine whether there is a preferable method to train individuals. In addition, intervention studies specifically tailored to adults to see whether improvements in literacy skills can be made past the formal school period in individuals with WS and investigate whether this generalises to further improvements in other areas of daily living. Longitudinal follow up of individuals will help to explore the age related differences in adaptive functioning. Integrating more information about the home environment, including things such as parental expectations and siblings' adaptive abilities will be useful in contribution to our knowledge to be able to make appropriate recommendations about how best to support families as they endeavour to assist their child develop independence in life skills.

Appendix

MACQUARIE UNIVERSITY

Research Office
Research Hub, Building C5C East
MACQUARIE UNIVERSITY NSW 2109

Phone **+61 (0)2 9850 8612**
Fax +61 (0)2 9850 4465
Email ro@vc.mq.edu.au

Ethics
Phone **+61 (0)2 9850 6848**
Email ethics.secretariat@vc.mq.edu.au

16 April 2009

Dr Melanie Porter
Department of Psychology
Faculty of Human Sciences

Reference: HE27FEB2009-R06353

Dear Dr Porter,

FINAL APPROVAL

Title of project: ***"Genes, brains and cognition: a systematic examination of the links with Williams syndrome"***

Thank you for your recent correspondence. Your response has addressed the issues raised by the Ethics Review Committee (Human Research) and you may now commence your research.

Please note the following standard requirements of approval:

1. Approval will be for a period of twelve (12) months. At the end of this period, if the project has been completed, abandoned, discontinued or not commenced for any reason, you are required to submit a Final Report on the project. If you complete the work earlier than you had planned you must submit a Final Report as soon as the work is completed. The Final Report is available at: http://www.research.mq.edu.au/researchers/ethics/human_ethics/forms

2. However, at the end of the 12 month period if the project is still current you should instead submit an application for renewal of the approval if the project has run for less than five (5) years. This form is available at http://www.research.mq.edu.au/researchers/ethics/human_ethics/forms. If the project has run for more than five (5) years you cannot renew approval for the project. You will need to complete and submit a Final Report (see Point 1 above) and submit a new application for the project. (The five year limit on renewal of approvals allows the Committee to fully re-review research in an environment where legislation, guidelines and requirements are continually changing, for example, new child protection and privacy laws).

3. Please remember the Committee must be notified of any alteration to the project.

4. You must notify the Committee immediately in the event of any adverse effects on participants or of any unforeseen events that might affect continued ethical acceptability of the project.

5. At all times you are responsible for the ethical conduct of your research in accordance with the guidelines established by the University http://www.research.mq.edu.au/researchers/ethics/human_ethics/policy

If you will be applying for or have applied for internal or external funding for the above project it is your responsibility to provide Macquarie University's Research Grants Officer with a copy of this letter as soon as possible. The Research Grants Officer will not inform external funding agencies that you have final approval for your project and funds will not be released until the Research Grants Officer has received a copy of this final approval letter.

Yours sincerely

p.p.

Ms Karolyn White
Director of Research Ethics
Chair, Ethics Review Committee (Human Research)