Health and social outcomes in adults with Williams syndrome: Findings from cross-sectional and longitudinal cohorts

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ABSTRACT

Previous studies have investigated trajectories of cognitive, language and adaptive functioning in Williams syndrome (WS) but little is known about how other aspects of the Williams syndrome behavioural phenotype change across the life-span. Therefore, the present study examined age associated changes in a number of different domains of functioning in adults with WS. Semi-structured interviews and standardized assessments of physical health, educational and occupational levels, self-care and independence, social interactions, and behavioural difficulties were conducted with adults with WS and their parents/carers. Ninety-two individuals with genetic confirmation of WS (50 males, 42 females) participated in a cross-sectional study (age range 19–55 years; mean 32 years) and 43 individuals (21 males, 22 females) were involved in a longitudinal study as they had taken part in a previous study of adults with WS. Not all individuals in the longitudinal cohort had genetically confirmed WS. Cross-sectional analyses revealed no age related differences in the areas of functioning investigated. However, in the longitudinal sample, significant improvements were reported in physical health, self-care and occupational skills. Social skills and adaptive behaviours had generally improved while behavioural difficulties had declined. In both cross-sectional and longitudinal cohorts there were significant correlations between adaptive functioning and IQ. These findings are consistent in indicating no age related decline in social or adaptive functioning in adults with WS, at least up to the age of 50–55 years.

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1. Introduction

Williams syndrome (WS) is a neurodevelopmental disorder caused by a micro-deletion of at least 25 genes on chromosome 7q11.23 (Hillier et al., 2003). Prevalence estimates are generally around 1 in 20,000 live births (Morris & Mervis, 1999). Much of the research on Williams Syndrome has focused on the associated physical health complications, many of which arise from the elastin insufficiency caused by the genetic deletion. These include renal abnormalities, cardiovascular problems such as supravalvular aortic stenosis, hypertension, gastrointestinal complications, urinary tract and joint problems (Pober & Morris, 2007). Cognitive development, too, is impaired, with most individuals having an IQ in the mild intellectual impairment range. The cognitive profile is also unusual, with relative strengths in areas

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such as expressive vocabulary, face processing, auditory rote memory and some aspects of social cognition, but weaknesses in skills relating to verbal comprehension, number, planning, problem solving and spatial cognition (Howlin, Elison, & Stinton, in press; Martens, Wilson, & Reutens, 2008). With respect to behavioural characteristics, individuals with Williams Syndrome typically display an excessive interest in other people, and lack of social inhibition and over friendliness can give rise to difficulties, especially in adulthood (Davies, Udwin, & Howlin, 1998). Comorbid behavioural and mental health problems include attention deficit hyperactivity disorder, generalized anxiety and affective disorders, specific phobias and problems with preoccupations and obsessions (e.g. Cherniske et al., 2004; Einfeld, Tonge, & Rees, 2001; Kennedy, Kaye, & Sadler, 2006; Leyfer, Woodruff-Borden, Klein-Tasman, Fricke, & Mervis, 2006; Stinton, Elison, & Howlin, 2010).

1.1. Background to present study

Although the physical and cognitive abnormalities associated with Williams Syndrome have been well studied, most research has involved children rather than adults; there have been few investigations of transition into adult life, or of how the characteristic behavioural patterns may change with age. In a previous study we reported on the health and social and behavioural problems experienced by a relatively large sample of individuals with Williams Syndrome when aged, on average, 26 years (range 19–39 years; Davies, Howlin, & Udwin, 1997; Howlin, Davies, & Udwin, 1998). That study identified high rates of physical, behavioural, social and emotional problems in the adults involved. A subsequent postal survey of over 200 families (mean age of individuals with WS = 30 years; Howlin & Udwin, 2006) also indicated significant levels of physical and behavioural difficulties; over two-thirds of the sample surveyed remained highly dependent on their families for support; educational and employment attainments were low and the quality of health care and general support was considered to be poor.

The present study examined physical health, self-care, educational and occupational outcomes, social interactions and independence, and behaviour problems in a larger, older sample of adults. Many of these individuals were involved in our earlier adult study (Davies et al., 1997; Howlin et al., 1998) although at that time not all had their diagnosis confirmed by FISH testing. The findings are thus divided into 2 separate sections: a cross-sectional study of adults aged 19–55 years, all of whom had their diagnosis genetically confirmed, and a longitudinal study of individuals involved in our previous study of adults, not all of whom had a genetically ascertained diagnosis. Information on cognitive outcomes and mental health problems in the total sample are reported elsewhere (Howlin et al., in press; Stinton et al., 2010).

The main aims of the study were to investigate age related differences and, in the longitudinal sample, changes over time, in health, living and work arrangements, independence and social functioning, and behavioural problems.

2. Materials and methods

2.1. Participants

2.1.1. Recruitment

Potential participants were identified via the UK Williams Syndrome Foundation database and parents of all individuals aged 18+ were contacted to seek their agreement to take part in the study. Inclusion in the cross-sectional study required that all participants had a diagnosis confirmed by FISH test. For inclusion in the longitudinal study, participants were required to have been involved in the previous study of adults conducted by Howlin et al. (1998).

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2.1.2. Participation rates

Cross-sectional sample: One hundred eight individuals fulfilled inclusion criteria. Eight families could not be contacted, 7 declined to take part and 1 adult with WS had died (unspecified cardiovascular disease) leaving a total of 92 participants (85% of potential cohort; 50 females, 42 males). The mean age was 32 years (sd 8.1; range 19.9–55.3 years); mean full scale IQ was 56.7 (sd 7.2; range 44–80). In order to examine age related differences in functioning the sample was divided into three age groups (19–29 years 11 months, N = 44; 30–39 years 11 months, N = 31, and 40 years+, N = 17).

Longitudinal sample: Seventy individuals had been involved in the previous adult study. Sixteen families could no longer be contacted, 1 adult subsequently received a negative FISH test result and 4 adults had died (2 cause unknown; 1 "natural causes"; 1 of septicaemia following kidney operation), leaving a total of 49 participants (70%; of original cohort). Parent/carer information was not available for 6 of these individuals (2 adults did not have a carer who knew them well enough to complete the interview, an additional adult had died before the interview could be conducted and 3 parents declined to be interviewed). Data on physical and social changes over time are thus based on 43 adults (21 males, 22 females). The average time elapsed since the adults had been seen previously was 12 years (mean age when first seen = 24.9 years (sd 5.5, range 18–37.4 years), mean age at follow-up = 37.3 years (sd 5.7, range 30–49.6 years). Full scale IQ at the initial assessment was 58.4 (sd 7.6, range 46–83). At follow-up mean IQ was 62.6 (sd 6.3,