



Behavioral features of Williams Beuren syndrome compared to Fragile X syndrome and subjects with intellectual disability without defined etiology

D. Pérez-García^a, R. Granero^b, F. Gallastegui^c, L.A. Pérez-Jurado^{c,d}, C. Brun-Gasca^{a,*}

^a Department of Clinical Psychology and Health Psychology, Faculty of Psychology, Universitat Autònoma de Barcelona, Edifici-B, 08193-Bellaterra (Cerdanyola del Vallès), Spain

^b Department of Psychobiology and Methodology of Health Sciences, Faculty of Psychology, Universitat Autònoma de Barcelona, Spain

^c Department of Experimental and Health Sciences, Universitat Pompeu Fabra, and Centro de Investigación Biomédica en Red de Enfermedades Raras (CIBERER), Barcelona, Spain

^d Program in Molecular Medicine and Genetics, Hospital Universitari Vall d'Hebron, Barcelona, Spain

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ABSTRACT

Williams-Beuren syndrome (WBS) is a genetically determined neurodevelopmental disorder caused by a heterozygous deletion of 26–28 genes on chromosome band 7q11.23. During the past few years, researchers and clinicians have significantly contributed to define the phenotype of the syndrome, including its cognitive and behavioral aspects. However, it is not well known yet whether the psychological problems are specific to the syndrome or secondary to the intellectual disability (ID). The aim of our study was to better define the psychopathological profile of WBS and whether or not it is related with IQ or anxiety symptoms. Twenty-five subjects (12 girls, 13 boys) with a diagnosis of WBS were compared to 27 boys with Fragile X Syndrome and to 24 boys with ID of non-specific etiology using the Child Behavior Checklist. Anxiety, depression and attention problems were the main behavioral problems found in WBS with no gender differences. Significant differences between cohorts were observed in somatic complaints, delinquent behavior, aggressive behavior, and externalizing problems. Some associations between IQ and anxiety items were found. The findings are discussed in terms of behavioral phenotypes, genetic implications and ID.

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1. Introduction

Williams-Beuren syndrome (WBS) is a neurodevelopmental disorder mainly characterized by dysmorphic facial features, vascular stenoses, abnormalities of calcium and glucose metabolism, and intellectual disability (ID) with visuospatial deficits and hyperacusis, caused by a heterozygous deletion of 26–28 genes on chromosome band 7q11.23 (Bayes, Magano, Rivera, Flores, & Perez Jurado, 2003). Multiple health problems can appear during childhood and continue to be a major area of concern for many parents, who also complain about the lack of adequate support and global care (Howlin & Udwin, 2006).

* Corresponding author. Tel.: +34 93 581 27 78.

E-mail address: Carme.Brun@uab.cat (C. Brun-Gasca).

The cognitive profile of WBS is characterized by some relative strengths and weaknesses: strength in auditory rote memory, social skills and in select aspects of language, and remarkable weakness in visuospatial and visuomotor skills (Mervis & Klein-Tasman, 2000). Standardized testing demonstrates a full-scale IQ averaging 50–60, indicative of mild-to-moderate ID in most cases, though IQs span from 40 to 100 (Martens, Wilson, & Reutens, 2008).

The concept of “behavioral phenotype” was introduced in 1972 by Nyhan in his presidential address to the Society for Pediatric Research. *Behavioral phenotype is a characteristic pattern of motor, cognitive, linguistic and social abnormalities which is consistently associated with a biological disorder. In some cases, the behavioral phenotype may constitute a psychiatric disorder; in others, behaviors which are not usually regarded as symptoms of psychiatric disorders may occur* (Flint & Yule, 1994). The definition of the behavioral phenotypes of each syndrome will lead to effective treatments and special preventive programs.

A distinctive personality profile with high sociability, empathy and excessive anxiety appears characteristic of WBS. Specifically, WBS children show similar extraversion and agreeableness scores as children attending regular schools but they score much higher on agreeableness than other children with ID, such as those with Prader Willi or Fragile X syndrome (FXS)s (Van Lieshout, De Meyer, Curfs, & Fryns, 1998). WBS children also score lower than non clinical children on consciousness, emotional stability, openness, motor activity and irritability, but with high mean ratings on shyness and empathy, composing a gregarious, people-oriented, tense, sensitive, and visible personality profile (Klein-Tasman & Mervis, 2003). This distinctive personality pattern changes with aging. At older ages, WBS individuals show lower extraversion, lower emotional stability and lower motor activity (Van Lieshout et al., 1998). In spite of their friendly personality, many WBS adults are socially isolated.

Children with WBS have higher rates of overall behavioral/emotional disturbance and are also more likely to be diagnosed as “psychiatrically disordered” than other children with ID (Einfeld, Tonge, & Florio, 1997). Studies show a higher prevalence of specific phobia and generalized anxiety disorder in comparison to the general population (Phillips & Klein-Tasman, 2009) or other children with intellectual disabilities (Klein-Tasman & Mervis, 2003). Other relevant problems, including sleep disturbances (Einfeld, Tonge, & Rees, 2001), communication disturbances (Einfeld et al., 1997), and attention deficit/hyperactivity disorder (ADHD) (Leyfer, Woodruff-Borden, Klein-Tasman, Fricke, & Mervis, 2006) have also been reported among persons with WBS.

Gender differences have been found in the prevalence of fears and hyperacusis, being higher in females (Blomberg, Rosander, & Andersson, 2006). Longitudinal studies also show that most problems persist over a period of 4 years and into adulthood (Einfeld et al., 2001), with some changes during aging. In particular, adults with WBS appear to be at increased risk, relative to children, for depressive disorder and generalized anxiety disorder, while Attention Deficit Hyperactivity Disorder (ADHD) might be more common, or cause more functional impairment in children than in adults (Dodd & Porter, 2009). Depressive disorder manifestations in WBS usually start at the age of 15–25 years (Dodd & Porter, 2009).

Anxiety is nowadays also considered a significant feature of the behavioral phenotype of WBS (Leyfer, Woodruff-Borden, & Mervis, 2009). Despite its importance, little is known about how to focus prevention programs and intervention. While IQ inversely correlates with anxiety in WBS according to some authors, indicating that intelligence may be a protective factor (Dimitropoulos, Ho, Klaiman, Koenig, & Schultz, 2009), other studies show no correlation (Woodruff-Borden, Kistler, Henderson, Crawford, & Mervis, 2010).

Specific criteria have been established for the diagnosis of psychopathological disorders in subjects with ID, always considering somatic complaints as possibly related features (Diagnostic Manual-Intellectual Disability, DM-ID) (Fletcher, Loschen, Stavrakaki, & First, 2007).

As summarized above, the research during the last few years has provided a much better definition of the clinical phenotype of WBS including its cognitive and behavioral aspects. There is also evidence for significant psychopathological problems along the life span but the psychopathology in WBS is not so well defined. It is not clear whether these traits and disorders are specific of the syndrome or secondary to the ID.

Therefore, the aim of our study was to contribute to a better definition of the behavioral features that may be specific of the WBS and its putative correlation with IQ, using FXS and non-specific ID as control groups.

2. Method

2.1. Sample

The first cohort were 25 individuals (12 girls, 13 boys) with a diagnosis of WBS confirmed by molecular genetics (existence of a heterozygous deletion at the 7q11.23 chromosomal band) aged 5–27 years (mean = 14.3, SD = 1.3) and IQ range 40–93 (mean = 55.4, SD = 2.8). All participants in the WBS group were part of a bigger research of medical aspects (EC07/0856 2008–2010) conducted at Hospital Vall d’Hebron of Barcelona with patients from all over Spain. Control groups were obtained from a previous study of patients with ID (Artigas & Brun, 2004). One group was composed of 27 boys with confirmed diagnosis of FXS (with *FMR1* gene mutation) aged 6–18 (mean = 12.4, SD = .71) and IQ range 35–89 (mean = 48.2, SD = 2.1). The second control group consisted on 24 boys with ID but non-specific etiology (IDNS) aged 5 to 18 (mean = 12.8, SD = .71) and IQ range 30–76 (mean = 49.2, SD = 2.6). Patients of these two groups were followed in the Genetic Service of the

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