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Evaluation of the influence of clinical symptoms on the quality of life in individuals with Williams-Beuren Syndrome

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Abstract: Introduction: Williams-Beuren Syndrome (WBS) results from a deletion in the chromosome 7q11.23. Psychiatric symptoms, such as Generalized Anxiety Disorder (GAD), Attention Deficit Hyperactivity Disorder (ADHD), and others, have been described in this syndrome. Objective: The main purpose of this study was to identify related clinical symptoms in individuals with WBS and the impact of this disorder in their quality of life. Method: The study sample included 42 individuals aged 11 to 16 years divided into two groups: a study group composed of 20 individuals with WBS and a control group comprising 22 individuals with typical development. The following instruments were used in this evaluation: Brazilian Criterion of Economic Classification (CCEB), to estimate the purchasing power of urban households; Strengths & Difficulties Questionnaire (SDQ), to provide a useful measure of psychopathology applied to children and youth aged 4-17 years; and Quality of Life Evaluation Scale (AUQEI), to assess the feeling of well-being according to the opinion of the child and adolescent. Results: The results suggest that adolescents with WBS have a good quality of life despite presenting greater possibilities of psychiatric symptoms. These results are valid considering that quality of life is measured from the opinion of the assessed individual. Conclusion: We hypothesized that adolescents with WBS can present a distortion of reality regarding the positive/optimistic factor, possibly based on some characteristics of the syndrome such as being always cheerful and smiling, which are factors associated with intellectual disability.

Keywords: *Williams-Beuren Syndrome, Clinical Symptoms, Quality of Life.*

Avaliação da influência dos sintomas clínicos na qualidade de vida de indivíduos com Síndrome de Williams-Beuren

Resumo: Introdução: A Síndrome de Williams-Beuren (SWB) é resultado da deleção do cromossomo 7q11.23. A presença de transtornos psiquiátricos, tais como Transtorno de Ansiedade Generalizada, Déficit de Atenção e Hiperatividade, entre outros, já foram descritos nesta síndrome. Objetivo: Este estudo teve como objetivo identificar possíveis sintomas clínicos nos indivíduos com SWB e as possíveis consequências na qualidade de vida. Método: O grupo amostral com SWB constituiu-se por 20 indivíduos com idade entre 11 e 16 anos e 22 indivíduos com desenvolvimento típico (DT). Os instrumentos utilizados foram: Critério de Classificação Econômica Brasil para estimar o poder de compra das famílias urbanas; o Questionário de Capacidades e Dificuldades (SDQ), que possibilita uma medida útil em psicopatologia aplicada a crianças e jovens com 4 a 17 anos de idade; e o Questionário de Avaliação de Qualidade de Vida em Crianças e Adolescentes (AUQEI), que tem como objetivo avaliar a sensação de bem-estar mediante a opinião da própria criança e adolescente. Resultados: Os resultados sugerem que os

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adolescentes com SWB possuem uma boa qualidade de vida, apesar de apresentarem maiores possibilidades de sintomas psiquiátricos. Estes resultados são válidos, já que a qualidade de vida é mensurada a partir da subjetividade do indivíduo avaliado. Conclusão: Chega-se à hipótese de que os adolescentes com SWB podem apresentar uma distorção da realidade para o fator positivo/otimista, possivelmente baseada em algumas características da própria síndrome, tais como: são sempre alegres e sorridentes, fatores associados à deficiência intelectual.

Palavras-chave: *Síndrome de Williams, Sintomas Clínicos, Qualidade de Vida.*

1 Introduction

Williams-Beuren Syndrome (SWB) was first described by Williams, Barratt-Boyes and Lowe (1961) and Beuren, Apitz and Harmjanz (1962) who observed typical facial features associated with aortic supravalvular stenosis, childhood hypercalcemia and intellectual disability in unrelated patients. It is a relatively rare disorder of neurological development, resulting from a hemizygous deletion of 25-30 genes on chromosome 7q11.23 (EWART et al., 1993; KORENBERG, 1993; HILLIER et al., 2003). SWB occurs in about 1 in 8,000 live births (MARTENS; WILSON; REUTENS, 2008).

This syndrome has greater potentialities in language abilities contrasted with the severe deficiency in visuospatial ability. In this way, it is often described by a profile of "peaks and valleys." The people with this syndrome are considered highly sociable, smiling and with a high level of verbal development. Frequently, they present mild to moderate intellectual deficiency and difficulties in spatial reasoning, problem-solving and fine motor skills (NUNES, 2010). Moreover, they exhibit a strong impulse to engage socially, with increased propensity to approach strangers (JARVINEN-PASLEY et al., 2008; JAVINEN; BELLUGI, 2013; HAAS; REISS, 2012).

The literature also describes the individuals with SWB as more anxious, distracted, hyperactive and prone to experiencing relationship difficulties with peers of the same chronological age (children of the same age group) or those with similar levels of cognitive impairment (DYKENS, 2003; EINFELD; TONGE; FLORIO, 1997; EINFELD; TONGE; REES, 2001).

Mervis et al. (2015) evaluated 79 individuals with SWB between 18 months and 17 years old and 12 adults. The results indicated very wide variations compared to standardized assessments of intelligence, vocabulary and academic performance. Levels of adaptation range from severe to medium deficiency, as well as, disruptive behavior have been classified since the extreme difficulty in their absence. More than half of the participants were diagnosed with anxiety disorder. Also, a quarter of the children were diagnosed with an oppositional defiant disorder or disruptive behavior disorders - without further specification, and a third of the children positively had an autism spectrum disorder.

A cross-sectional study analyzed the adaptive behavior profile in 100 adolescents and adults with SWB and used the Independent Revised - Behavior (SIB-R) scale. The findings indicated a significant deficit in motor skills and community life skills compared to performance in social interactions, communication skills, and personal life skills (FU, 2015). Malone and Stoneman (1990) and Beadle-Brown et al. (2000) emphasize that the development of cognition is associated with the development of adaptive behavior and, consequently, influencing the quality of life. Occupational therapy works on the common mental health disorders of childhood and adolescence and aims to improve health, promote autonomy and social inclusion (FLEITLICH-BILYK; GOODMAN, 2004).

A Brazilian study (DEL COLE et al., 2013) analyzed the occupational performance in the Basic Activities of Daily Living (ABVD) and Instrumental Activities of Daily Living (AIVD) in 10 adolescents and young adults with SWB and identified that nine out of ten individuals have severe dependence alluding to family care in ABDV and AIVD. The main behavioral problems reported by the parents were the lack of attention, change in social contact and excessive anxiety. Also, this study found difficulties in interactions and personal relationships, significant impoverishment of the social repertoire and occupational roles, in the social/environmental context in which they were.

Studies show the quality of life as in possessing the resources necessary to satisfy individual needs and desires, participating in activities that allow for personal development, self-realization, and a satisfactory comparison between oneself and others (DANTAS, 1997; SHIN; JOHNSON, 1978; HINDS, 1990). For children and adolescents, the quality of life perspective is also associated with the distance between their desires and the momentary reality (CALMAN, 1984). Mental health research focuses on issues that poor quality of life may be a consequence of dysfunction, inability, and disability (JANSEN et al., 2011).

From the perspective of the cognitive deficit, physical disability and clinical symptoms can affect the quality of life; the hypothesis is the perception of adolescents with SWB on their quality of life is inferior to adolescents with typical development (DT). Thus, the main objective of this study was to evaluate if the quality of life of adolescents with SWB is deficient in the adolescents with DT and if the presence of clinical symptoms negatively influences their quality of life.

2 Method

2.1 Sample group

The sample group with SWB was composed of 20 adolescents and the inclusion criteria were being chronological age between 11 and 16 years old, having the diagnosis of SWB genetically confirmed by clinical analysis and molecular cytogenetics (Fluorescence In Situ Hybridization - FISH), living in the State of São Paulo and being registered with the Brazilian Association of Williams Syndrome (ABSW).

The DT group consisted of 22 adolescents who were relatives or friends of the SWB group. The inclusion criteria established were living in the State of São Paulo and having a pairing with the SWB group in the requirements of age, gender, and economic classification.

2.2 Evaluation instruments

2.2.1 Brazil's Economic Classification Criteria (2011)

The questionnaire used was developed by the Brazilian Association of Companies in Research (ASSOCIAÇÃO ..., 2011), in which the criterion of Economic Classification Brazil emphasizes the function of estimating the purchasing power of urban families and the market division is defined exclusively by classes. It was constructed using statistical techniques that are always based on collectives, so the probability of correct classification is high, and the probability of error is low. Thus, incorrectly classified cases will be few so as not to distort the results of the investigation significantly.

2.2.2 Capacities and Difficulties Questionnaire (SDQ)

The SDQ is a brief questionnaire to monitor possible psychiatric symptoms in children and adolescents between 4 and 17 years old. The version selected for this study was P4-17 - SDQ and supplemental impact for parents of children or adolescents. The 25 SDQ items are divided into five diagnostic profiles: emotional symptoms (5 items); behavior problems (5 items); hyperactivity/lack of attention (5 items); relationship problem with colleagues (5 items); pro-social behavior (5 items).

The general score of difficulties is generated by the sum of the results of all the scales, except the pro-social behavior scale. The result can vary from 0 to 40 points, and possible clinical symptoms are present if at least 12 of the 20 points are identified as characteristic of the subject evaluated. The SDQ questionnaire indicates positive validity and reliability indices in 21 countries, including Brazil, characterizing its cross-cultural reach and its applicability in the child and adolescent mental health area (SAUR; LOUREIRO, 2012).

2.2.3 Evaluation Questionnaire of Quality of Life in Children and Adolescents (AUQEI)

The validated version of the "Child Quality of Life Scale" (in French, *Autoquestionnaire Qualité de Vie Enfant Imagé* - AUQEI), developed by Manificat and Dazord (1997), is an instrument that aims to evaluate the subjective feeling of well-being of the individual and part of the premise that the developing individual can express himself as to his subjectivity. The AUQEI scale is composed of 26 questions that explore family, social relationships, activities, health, bodily functions, and separation.

It is a self-assessment using the support of images, in which the adolescent responds to each question through the help of faces that express different emotional states (Figure 1). The adolescent is instructed to indicate, without definite time, the response that corresponds most to his feeling, in the proposed domain. However, it is firstly requested that the adolescent reports his experience with each of the alternatives, allowing him to better understand situations by presenting his experiences. The scale allows obtaining a profile of satisfaction of the individual before different situations (ASSUMPÇÃO et al., 2000).

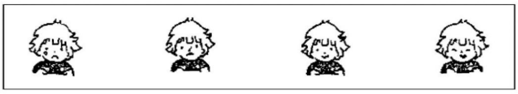

Figure 1. Faces of the Questionnaire for the Evaluation of Quality of Life in Children and Adolescents (AUQEI). Note: Feeling of faces starting from left to right: very sad, sad, happy and very happy.

This instrument has a step before the application of the questions to ensure the child or adolescent expressing his feelings to each of the options of answers. For the answer representing "very happy," the child is induced to remember some previous situation he had such feeling to better understand the options (ASSUMPÇÃO et al., 2000). Moreover, since this instrument has unclear images about the differences between them, it was necessary the researcher repeated in all the questions what the adolescent felt, observing the image indicated and corresponding during the evaluation process by caution, such as when he was asked: "When your parents talk about you, how do you feel?", "Very happy, happy, sad or very sad" (the researcher points the corresponding image).

AUQEI showed satisfactory psychometric properties and ease of use. There were 353 schoolchildren evaluated, aged four to 12 years old from the city of São Paulo, SP to validate this instrument for the Portuguese Language. A cut-off point of 48 was established for the general population, and the quality of life was considered unsatisfactory below this score. The study presented internal validity, with Cronbach's coefficient of 0.71 and indicated the adequate reliability of the instrument (ASSUMPÇÃO et al., 2000).

2.2.4 Wechsler Intelligence Scale for Children-third edition (WISC-III)

The WISC-III questionnaire is an instrument evaluating the development of some cognitive functions of children and adolescents aged 6 to 16 years old (WECHSLER, 1991). The estimated intelligence quotient (IQ) was calculated using the Cube and Vocabulary subtests. These subtests were chosen because they presented psychometric characteristics that correlate with total IQ (MELLO et al., 2011). The analysis showed that the adapted items presented adequate psychometric characteristics that allow the use of the instrument as a reliable means of diagnosis (FIGUEIREDO et al., 2008).

2.2.5 Vineland Adaptive Behavior Scale (VABS)

The VABS scale measures adaptive behavior, important data to aid the diagnosis of intellectual disability (DSM-5). This scale evaluates the individual´s ability to cope with environmental changes, learning how to perform everyday activities, and demonstrating independence in developing them (SPARROW et al., 1984). The scale is based on a structured interview with the primary caregiver. The evaluation time was approximately 25 to 90 minutes. VABS is organized into a framework with three main domains: communication, activities of daily living and socialization. The crude results obtained from the domains were weighted to fit according to chronological age, according to the VABS manual, and normalized to obtain a common metric (SPARROW et al., 1984). The psychometric data of the VABS scale are reliable and have been described in their manual through evidence of construct, content and criterion validity (SPARROW et al., 1984).

2.3 Procedure

The data were collected during 2011 and 2012 in the Psychiatry Department of the Federal University of São Paulo (UNIFESP), in reserved environments and at events held by ABSW in the city of São Paulo. A meeting was held with the family, and the adolescent for the data collection and the initial contact was carried out as follows: one part was by telephone and another, personally during the ABSW events.

After clarifying, understanding and agreeing on the purpose of this study and the evaluation procedures that would be applied, the parents authorized the participation of the children, signing the Informed Consent Form and answered, with a trained applicator, the following instruments: Brazil´s Economic Classification Criteria, SDQ, and the VABS scale.

The adolescents were first clarified about the evaluations and, after agreeing to participate, they signed the Term of Assent. Then, the AUQEI questionnaire and the WISC-III was answered by a trained applicator.

The duration of application of the Brazil´s Economic Classification Criteria, SDQ and VABS was 1 hour and the application of the AUQEI and WISC-III questionnaires lasted about 40 minutes.

2.4 Data analysis

Student's t-test was used for independent samples, using software R to compare the groups in each of the variables of interest.

Descriptive analysis of the data presented included mean and standard deviation. For all statistical procedures, the confidence interval was 95% ($p < 0.05$).

3 Results

3.1 Sample characteristics: sociodemographic data

There was no statistical difference between SWB and DT groups in the genders ($p>0.287$), age ($p>0.892$) and social class ($p>0.887$). However, as expected, there was a statistically significant difference for the estimated QI, for all domains of adaptive behavior and the total of the VABS scale. Thus, the DT group presented better performance about the SWB group in the estimated IQ ($p<0.001$) and in all domains of the VABS scale ($p<0.001$).

However, the effect size between the adaptive behavior performance averages between the DT groups and the SWB group was classified as a mean size effect for the socialization domain and classified as a large size effect for the communication domains, activities of daily life and total adaptive behavior. These data are described in Tables 1 and 2.

3.2 Quality of life and clinical data

There was no statistically significant difference between the SWB group (mean = 52.50 and SD = 8.19) and the DT group (mean = 51.41 and SD = 5.16, $p>0.613$) about quality of life as expected. Thus, the group with SWB had an above-average point than the DT group (Figure 2).

The SWB group (mean = 19.29 and SD = 4.12) had greater possibilities of clinical symptoms when compared to the DT group (mean = 8.05 and SD = 6.15, $p<0.001$). The results for the sub-domains of the SDQ scale were adolescents with SWB scored lower than the DT group in the sub-domain of "prosocial behavior" ($p<0.002$) (Figure 3). As expected, the SWB group presented greater possibilities for hyperactivity ($p<0.001$) (Figure 4), problems with colleagues ($p<0.001$) (Figure 5), emotional symptoms ($p<0.001$) (Figure 6), and behavior problems ($p<0.05$) (Figure 7).

4 Discussion

This study showed that there was no difference in the quality of life between the SWB group and the DT group. However, although the possible clinical symptoms are evident in the SWB group, such symptoms did not interfere with the quality of life of these adolescents.

Table 1. Sociodemographic data Table.

	Gender			Age		Economic Classification			WISC (QI)	
	Fem. N (%)	Male N (%)	p-value	Mean (SD)	p-value	A/B	C/D	p-value	Mean (SD)	p-value
DT	9 (41)	13 (59)	-	13.6 (1.8)	-	12 (54.5)	10 (45.5)	-	105.62 (16.77)	-
SWB	11 (55)	9 (45)	0.287	13.7 (1.8)	0.892	11 (52.4)	9 (47.6)	0.887	59.39 (9.07)	<.001**

Note: Typical development = DT; Williams-Beuren syndrome = SWB; Female = Fem.; Male. Social classes A/B and Social classes C/D. ** = Statically significant.

Table 2. Effect size of the mean adaptive behavior in the groups DT and SWB.

	1 DT Mean (SD)	2 SWB Mean (SD)	HC vs. WBS Cohen's d/effect size r (p)
lization	112.45 (17.40)	60.30 (26.09)	2.35/0.76* (*p*<.001)
Communication	103.66 (13.57)	40.82 (18.95)	3.82/0.88**(*p*<.001)
Activities of Daily Life	103.33 (18.89)	33.91 (15.57)	4.06/0.89**(*p*<.001)
Total Adaptive Behavior	108.62 (16.42)	41.39 (15.39)	3.60/0.87**(*p*<.001)

Note: Typical development = DT; Williams-Beuren syndrome = SWB; Socialization, communication and activities of daily living, and total adaptive behavior of the Vineland Scale. * medium size effect; ** large size effect.

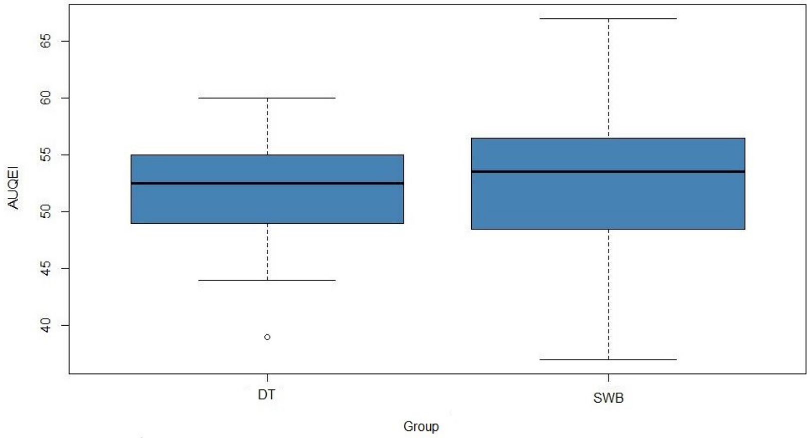

Figure 2. Distribution of the variable 'AUQEI' in groups with SWB and DT. Note: There was no statistical difference regarding the quality of life between the SWB group and the DT group.

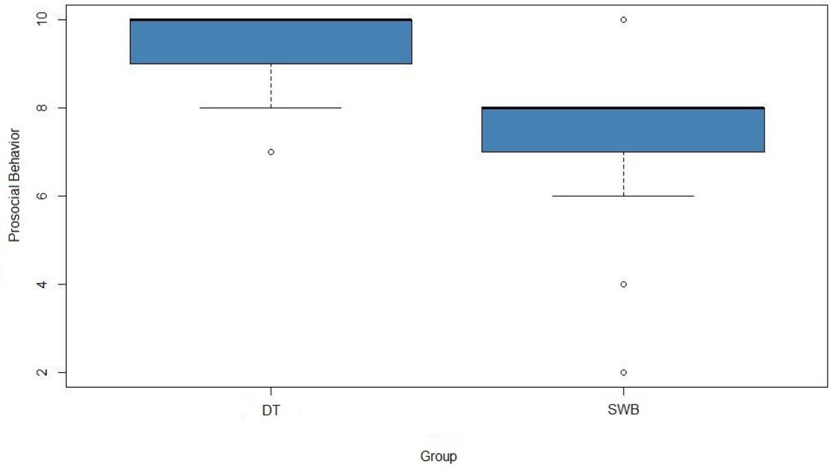

Figure 3. Distribution of the subdomain 'pro-social behavior' in SWB and DT groups. Note: Individuals with SWB had a lower score than the DT group in the "prosocial behavior" variable.

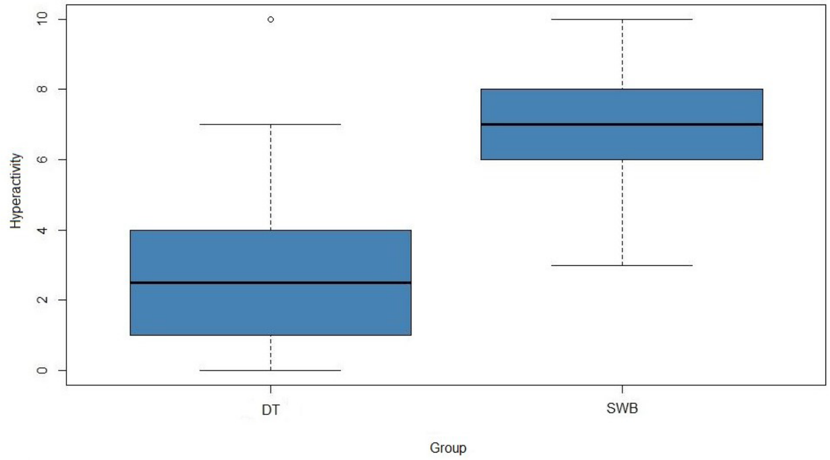

Figure 4. Distribution of subdomain 'hyperactivity' in SWB and DT groups. Note: Individuals with SWB had a higher score than the DT group in the "hyperactivity" variable.

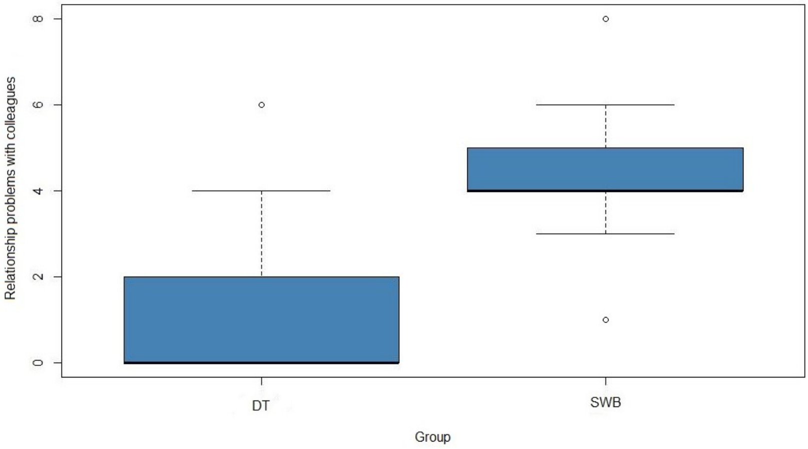

Figure 5. Distribution of subdomain 'peer relationship problems' in SWB and DT groups. Note: Individuals with SWB had a higher score than the DT group in the variable "peer relationship problems".

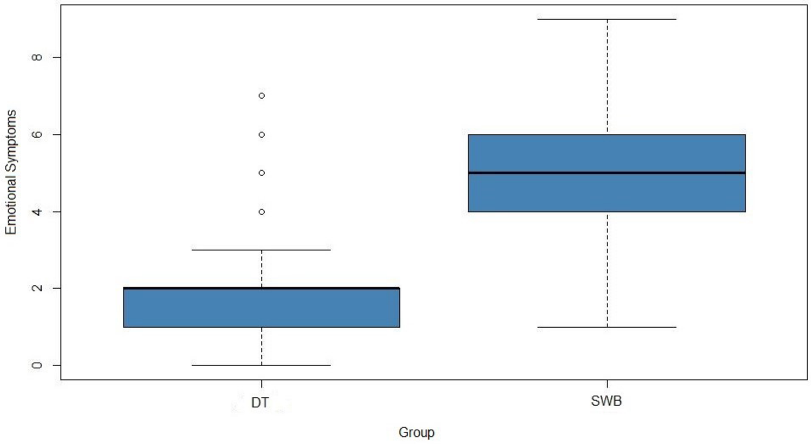

Figure 6. Distribution of subdomain 'emotional symptoms' in SWB and DT groups. Note: Individuals with SWB had a higher score than the DT group in the "emotional symptoms" variable.

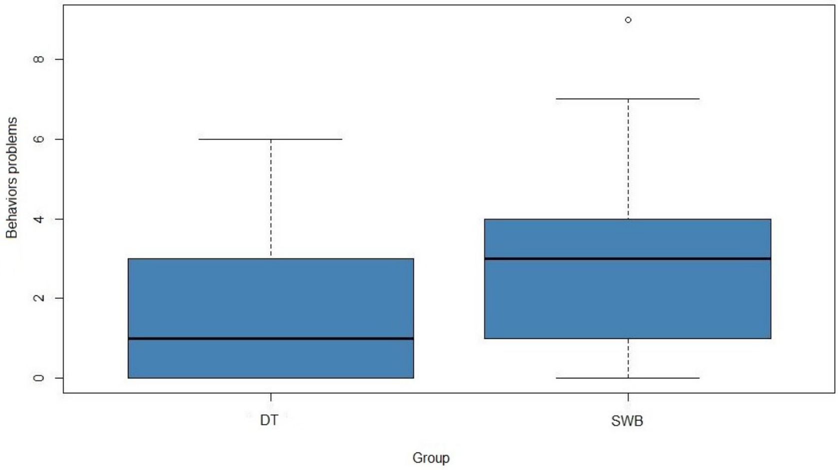

Figure 7. Distribution of the subdomain 'conduct problems' in SWB and DT groups. Note: Individuals with SWB had a higher score than the DT group in the variable "conduct problems" variable.

The findings related to clinical symptoms corroborate with the literature, since they report that individuals with SWB are more anxious, distracted, hyperactive and more likely to have difficulties of relationship with colleagues of the same chronological age, or with those at the same levels in cognitive deficits (EINFELD; TONGE; FLORIO, 1997; GREER et al., 1997).

Kennedy, Kaye and Sadler (2006) reported that 43% of a sample of 20 individuals with SWB met diagnostic criteria for attention-deficit/hyperactivity disorder (ADHD). Leyfer et al. (2006) also showed that 65% of 119 individuals with SWB were diagnosed with ADHD, as well as 54% for specific phobia.

According to Freeman et al. (2010), individuals with SWB and their families are aware of the difficulties regarding emotional and hyperactivity symptoms. However, when it comes to problems of peer relationships and prosocial behavior, they have difficulty in perceiving these deficits. Moreover, Gosh and Pankau (1994, 1997) suggest that although SWB is characterized by highly sociable behavior, these individuals show serious problems of social adaptation, including difficulties in developing and relating to colleagues.

The quality of life is a comprehensive concept, affected in a complex way by the physical health, psychological state, level of independence, social relationships and characteristics of the environment in which the individual lives (WORLD..., 1993). Thus, although the results obtained in this study suggest that adolescents with SWB had a quality of life classified in their subjective opinion as ‘good’ it is somewhat contradictory to say that the mental health issues observed in this study do not exert any influence.

However, one study showed that children and adolescents diagnosed with ADHD presented significant impairment in several domains associated with quality of life, since this diagnosis is associated with emotional and behavioral problems and, consequently, these problems had repercussions in their parents, causing them emotional suffering and interfering with family activities (KLASSEN; MILLER; FINE, 2006). Another study shows that children with ADHD have difficulties in generating appropriate (adaptive) responses at home, at school and in the community (DUNN; BENNETT, 2012) because they differ significantly from typical children in the sensory responses (DUNN; BENNETT, 2012; SHIMIZU; BUENO; MIRANDA, 2014). John and Mervis (2010) demonstrated that most children with SWB who had poor adaptive and executive functioning presented greater difficulties of sensorial modulation and problems of behaviors. Also, Amaral et al. (2013) observed that individuals with SWB of school age stand out for the friendly personality and good narrative. However, these children show high-stress index; and that stress is usually the consequence of an inadequate optimistic personality, as it gives rise to unpleasant consequences causing stress since they usually deal inadequately with certain social circumstances.

However, considering the World Health Organization’s definition of quality of life (THE WHOQOL..., 1995) as an individual’s perception of their position in life in the context of the culture and value systems in which they live and in relation to their goals, expectations, standards and concerns, it is suggested that the results of this study are valid, although the SWB group may have a lack of understanding about its limitations regarding social relationships, as pointed out by Freeman et al. (2010). Alternatively, the distortion of the understanding of the difficulties in maintaining social relationships, and the real perception of life is probably because these individuals are intellectually deficient (MARTENS; WILSON; REUTENS, 2008; MERVIS et al., 2000).

The AUQEI test proposes to qualify the quality of life through the subjectivity of the evaluated individual, so the results of this research are valid since the adolescents with SWB could respond to the questionnaire. Thus, it is suggested that disability or deficits cannot always be predicted to interfere with quality of life, as well as the possible clinical symptoms were not enough to show that the well-being of these adolescents would be compromised.

Also, AUQEI is widely used in investigations that seek to assess the quality of life in healthy children with some disorder, as in a study that showed similar quality of life indices in 20 children aged 4 to 12 years old with autism spectrum disorder, compared to 20 normal children, matched by gender and age (ELIAS; ASSUMPÇÃO JÚNIOR, 2006).

The last point to be discussed is the intervention in occupational therapy contributing to the development of individuals with SWB, since they present difficulties in several areas that are part of the scope of this profession, such as nonverbal reasoning, visuospatial perception, spatial representation, working memory, motor planning and executive functions (NUNES et al., 2013). In their study, Santos et al. (2015) observed that the motor focus intervention allows positive advances in the areas of fine motor, balance and spatial organization in individuals with SWB.

Thus, in agreement with the objectives of the intervention and demand of each subject, different approaches and resources can be used by occupational therapists to improve the functionality and performance of these individuals in their daily lives. Among some intervention techniques, the Sensory Integration therapy developed by the occupational therapist and neuroscientist Jean Ayres (1972) may be one of the options, since it aims to result in an adaptive response to the stimuli experienced by the individual (SHIMIZU; MIRANDA, 2012). However, further studies are needed to verify its efficacy and effectiveness in this population.

The first limitation of this study was the sample size given for convenience. After all, all adolescents enrolled in ABSW who met the inclusion criteria and who agreed to participate in this study were evaluated. However, the sample was limited to adolescents residing in the State of São Paulo. Therefore, it would be necessary to expand the sample size and apply the evaluations in other Brazilian regions. The second limitation was that there was no correlation that other outcomes could interfere with the quality of life of these adolescents, such as the adaptive behavior. Finally, there is little literature describing evaluations or interventions of occupational therapy in SWB, hindering to elaborate the discussion in this scope.

5 Conclusion

Therefore, in their perception of the world, adolescents with SWB are satisfied with the type of quality of life they lead in all segments of society, regardless of clinical symptoms that could interfere in a negative way in this quality. Thus, it is suggested that individuals with SWB may probably present a distortion of reality to the positive/optimistic factor, possibly based on one of the behavioral characteristics of the syndrome associated with intellectual disability.

Given the aspects investigated, Occupational Therapy professionals and other members of the multi/inter/transdisciplinary team are proposed to use the methodology applied in this study to evaluate and enrich the work with individuals with SWB, since they understood the issues Quality of life protocol and answered the questions in an appropriate way. Since this syndrome has been little touched by the area of occupational therapy, new studies could be carried out to provide more information, developing different methodologies, among others, contributing to all involved, and consequently, improving and enriching the methods and objectives of SWB interventions and also collaborating with the literature.

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Author's Contributions

Luísa carried out the evaluations cited in this study, analyzed and assisted in the conception of the text. Andrea guided the research project financed by FAPESP and carried out a review of the article. Rosana Aparecida contributed to the analysis and review of the text. Carolina supervised the evaluations carried out by Luísa and assisted in the analysis, the writing and the review of the text. All authors approved the final version of the text.

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