PHONOLOGICAL AWARENESS ASSESSMENT IN WILLIAMS SYNDROME

*Avaliação da consciência fonológica na S*índrome de *Williams*

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ABSTRACT

Purposes: assess the performance of children and adolescents with Williams Syndrome in tasks of phonological awareness and analyze it in terms of age, education and indicators of intellectual skills. **Methods**: twenty two children and adolescents (11 boys and 11 girls), aged 7 to 18 years old, took Block Design and Vocabulary subtests of Wechsler Intelligence Scale for Children to estimate intellectual skills, and the Phonological Awareness Test by Oral Production. **Results**: participants obtained estimate IQ values compatible with intellectual disability. Phonological awareness results indicated that participants with the syndrome had a lower performance in nine out of ten subtests of the Phonological Awareness Test compared to standard scores for children with same age and education level. No significant correlations were found among phonological awareness, intellectual skill indicators, age and level of education. **Conclusions:** results corroborate the findings of international researches that suggest difficulties in phonological awareness processes in Williams Syndrome. Considering the relevance of these findings, it is necessary to include continuous stimulation programs, including early intervention for preschoolers with Williams Syndrome, adaptation of teaching methods and curriculum.

KEYWORDS: Williams Syndrome; Intellectual Disability; Awareness

INTRODUCTION

Williams Syndorme (WS) is a genetic disorder caused by a multigenic hemizygous deletion in chromosome 7q11.23 that, in 95% of the cases, encompasses from 1,5 to 1,6 Mb (deletion of approximately 26 genes), with prevalence above 1:7.500 live borns[1-4]. WS is diagnosed through clinical assessment, usually during childhood, based on phenotypic traits such as prominent cheek, upturned nose, long philtrum and cardiovascular symptoms, especially supravalvular aortic stenosis. Other typical alterations are neurobehavioral dysfunctions, growth alterations, gastrointestinal and renal dysfunctions[5-7]. The definite diagnosis must be confirmed by genetic-molecular exams. Although there are more severe cases, most people with WS present mild to moderate intellectual disability (ID)[8].

In the last 10 years, the WS has been receiving considerable attention due to its specific cognitive profile[9-13]. In terms of neurocognitive phenotype, WS is often described as a profile of "peaks and valleys". People with WS are relatively proficient in concrete language skills and have a better performance in expressive language than in receptive; this contrasts sharply with the difficulties in syntactic-pragmatic language skills, structural and functional linguistic limitations, deficits in executive and visuospatial skills (work memory and planning) and severely impaired learning[12-14].

Studies on phonological awareness in individuals with WS have shown that the phonological process plays an important role for the acquisition of reading skills[15-17]. Phonological awareness is an intentional reflection on speech that has different levels, i.e., the segmentation of spoken language can happen in different units: a sentence can be segmented in words, words can be segmented in syllables

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and syllables can be segmented in phonemes[18,19]. Indeed, knowing that the language has a specific system of sounds, that these units are repeated in different spoken words and that orthography can be converted in phonology is a relevant aspect for reading acquisition. Thus, phonological processing is required for the acquisition of reading skills through decoding letters and groups of letters in their corresponding sounds. This skill is, in its turn, key to the capacity of reading words fluently[19-21].

Considering the relevance of phonological awareness for the acquisition of written language and the scarce number of studies that assess phonological awareness in children with WS, the present study had the objective of assessing the performance of children and adolescents with WS in phonological awareness tests and analyzing this performance in terms of age, education level and intellectual ability (estimate intelligence quotient - IQ).

METHODS

The present study was approved by the Ethics Committee of Universidade Presbiteriana Mackenzie and was registered under CEP/UPM nº 1191/11/2009 and CAAE nº 0090.0.272.000.09. According to ethical norms for researches with humans, all participants and their legal guardians were instructed in advance about the objectives of the study and signed an informed consent form.

Participants

22 children and adolescents diagnosed with WS took part of this study, 11 of them (50%) were male and 11 (50%) female, ranging from 7 to 18 years old (M=11,6; SD=3,7). Concerning school attendance, 12 (54,6%) are enrolled in a mainstream public school, 5 (22,7%) in private schools and 5 (22,7%) special education schools. Each child was assessed individually in a 90'-session with intervals between tasks.

Tools and Procedures

To assess the sample's intellectual potential we used Block Design and Vocabulary subtests of Wechsler Intelligence Scales, WISC-III[22] or WAIS-III[23] depending on the age, in order to obtain the estimate IQ[24].

After that, participants took the Phonological Awareness Test by Oral Production (PAT-OP) [19], which consists of ten subtests with 4 items each. Initially, the examiner presented two examples in order to explain what the child was expected to do:

1) syllabic synthesis: the child has to unite the syllables spoken by the examiner and speak the resulting word;
2) phonemic synthesis: the child has to join phonemes spoken by the examiner;
3) rhyme: the child has to choose, out of three words, two that end with the same sound;
4) alliteration: the child has to choose, out of three words, two that start with the same sound;
5) syllabic segmentation: the child has to divide a word spoken by the examiner into its component syllables;
6) phonemic segmentation: the child has to divide a word spoken by the examiner into its component phonemes;
7) syllabic manipulation: the child has to add and subtract syllables and say the word that is formed;
8) phonemic manipulation: the child has to add or subtract phonemes and say the word that is formed;
9) syllabic transposition: the child as to invert syllables and say the word that is formed;
10) phonemic transposition: the child has to invert phonemes and say the word that is formed.

In order to assess the response patterns for each participant, the total number of correct answers in the test was analyzed. Individual results were then compared to standardized data[25] obtained from the performance of children enrolled in pre-school and elementary school, with conversion of raw scores into standard scores (Mean = 100; Standard Deviation = 15). We used Wilcoxon analysis to compare mean results according to level of education (WS group X Normative group) and Pearson correlation analysis to describe associations among age, education and indicators of intellectual abilities. Results with p values ≤0,05 were considered significant.

RESULTS

Table 1 presents the general profile of the 22 children and adolescents with WS in terms of gender, age, education, intellectual abilities (estimate IQ) and total performance at the PAT-OP. Children and adolescents that formed the sample obtained estimate IQ values compatible with mild to moderate intellectual disability according to the standardization of the test.

The comparison of PAT-OP results of children with WS and normative data according to age[25] revealed that 3 participants had "low" classification while 19 were classified as "very low". However, considering that most individuals with WS do not study at the grade corresponding to their age, we have opted to consider the mean age equivalent

to each grade. Thus, we considered the following: 7 years old = 1st grade; 8 years old = 2nd grade; 9 years old = 3rd grade; 10 years old = 4th grade; 11 years old = 5th grade; 12 years old = 6th grade; 13 years old = 7th grade; and 14 years old = 8th grade. Then, we proceeded to comparing normative data according to grade, so that participants with WS were compared to younger children but which study at the same grade. Concerning the classification at PAT-OP, the comparison of performances of children with WS with normative data according to education[25] revealed that one participant presented low classification while the remaining 21 presented "very low" classification. These findings indicate that children with WS had performances below the expected at PAT-OP considering standard scores according to age and level of education, even when compared to younger children. Table 1 shows the classification of participants according to age and level of education.

Special education (SE) students were compared to the grade that corresponds to their age: 8y10m with 1st grade; 11y with 5th grade; 13y7m with 7th grade; and above 14y with 8th grade.

Table 2 shows that phonemic aspects of phonological awareness present a higher degree of difficulty in syllabic aspects.

In the studied sample, the group presented relatively high percentages of aspects of syllabic synthesis, rhyme, alliteration and syllabic segmentation; unlike aspects of phonemic synthesis, segmentation, manipulation and transposition, which were relatively low.

Table 3 summarizes results of the group with WS at PAT-OP for each school grade. Wilcoxon analysis revealed a significant difference ($Z = -4{,}110$; $p < 0{,}001$) between the performance of the WS group and the expected result according to level of education based on PAT-OP's normative data[25].

A Pearson correlation analysis was also made of total performance at PAT-OP and age (months), years of study / education (1st to 8th) and indicators of intellectual abilities. As it can be seen on Table 4, no significant correlations were observed.

Table 1 – Characterization of participants, including gender, education, IQ, total score at pat-op and classification considering normative data

N	Gender	Age	Grade	IQ	Total PAT-OP	Classification PAT-OP (Age)	Classification PAT-OP (Education)
1	F	7y 5m	1st	77	09,5	Low	Very low
2	M	7y 9m	1st	62	11,0	Low	Very low
3	F	8y	1st	83	05,5	Very low	Very low
4	M	9y1m	1st	56	13,5	Very low	Very low
5	M	16y3m	1st	45	11,0	Very low	Very low
6	F	8y10m	1st SE	80	12,5	Very low	Very low
7	M	8y 1m	2nd	68	06,5	Very low	Very low
8	M	9y2m	2nd	85	22,0	Low	Low
9	M	12y10m	2nd	49	08,0	Very low	Very low
10	F	17y10m	2nd	48	09,0	Very low	Very low
11	M	9y9m	3rd	65	11,5	Very low	Very low
12	F	10y10m	3rd	56	16,0	Very low	Very low
13	F	14y	3rd	68	19,0	Very low	Very low
14	F	11y2m	4th	54	06,0	Very low	Very low
15	F	14y2m	4th	62	18,5	Very low	Very low
16	M	12y11m	5th	68	21,5	Very low	Very low
17	F	11y	5th SE	59	11,0	Very low	Very low
18	M	15y	6th	56	19,0	Very low	Very low
19	M	16y10m	6th	56	06,0	Very low	Very low
20	F	13y7m	7th SE	45	04,0	Very low	Very low
21	M	17y11m	8th SE	65	08,0	Very low	Very low
22	F	18y3m	8th SE	62	11,0	Very low	Very low

N: Participant number; IQ: Intelligence Quotient; PAT-OP: Phonological Awareness Test by Oral Performance; M: Male; F: Female; SE: Special Education.

Table 2 – Descriptive analysis of phonological awareness aspects

Aspect	N	Mean	SD	Minimum	Maximum
Syllabic synthesis	22	3,8	0,4	3	4
Phonemic synthesis	22	1,0	0,9	0	3
Rhyme	22	0,9	1,2	0	4
Alliteration	22	1,0	1,3	0	4
Syllabic segmentation	22	3,0	1,4	0	4
Phonemic segmentation	22	0,5	0,8	0	3
Syllabic manipulation	22	1,2	0,9	0	3
Phonemic manipulation	22	0,3	0,6	0	2
Syllabic transpositions	22	0,3	0,8	0	3
Phonemic transposition	22	0,1	0,3	0	1

N: Number of participants; SD: Standard deviation.

Table 3 – Mean total correct answers in pat-op for the group with Williams Syndrome and normative group

PAT-OP	Grade	N	Mean age	WS	Expected for grade
Total PAT-OP	1st grade	6	9y1m	10,50 ± 2,8	12
	2nd grade	4	11y6m	11,38 ± 7,1	16
	3rd grade	3	11y3m	15,50 ± 3,7	22
	4th grade	2	12y7m	12,30 ± 8,8	26
	5th grade	2	11y5m	16,25 ± 7,4	29
	6th grade	2	15y5m	12,50 ± 9,1	27
	7th grade	1	13y7m	4,00 ± 0,0	32
	8th grade	2	17y7m	9,50 ± 2,1	32

Special education (SE) students were compared to the corresponding grade for their age: 8y10m with 1st grade; 11y with 5th grade and 13y7m with 7th grade and above 14y with 8th grade.
N: Number of participants per grade; PAT-OP: Phonological Awareness Test by Oral Production; WS: Group with Williams Syndrome.

Table 4 – Pearson correlation analysis of results in the phonological awareness test and factors age (months), education (1st to 8th grade) and indicators of intellectual skills

		Age	Education	IQ
TOTAL PAT-OP	*r*	-0,011	-0,06	0,294
	p	0,962	0,801	0,184
	N	22	22	22

N: Number of participants; IQ: Intelligence quotient; PAT-OP: Phonological Awareness Test by Oral Production; p: significance; r: correlation coefficient.

DISCUSSION

Compared to normative data, results obtained by individuals with WS were lower in almost all PAT-OP subtests. Only at the syllabic synthesis subtest did individuals with WS present adequate results in relation to normative data indexes. This information suggests individuals with WS acquire syllabic awareness before phonemic awareness, as it is the case for people with typical development[18]. Moreover, syllabic skills seem to be less impaired than general intelligence skills, as shown in Table 1.

However, it is important to point that this is the simplest subtest, seeing that syllabic analysis and other supra-segmental skills tend to develop more naturally once that syllables are units that require less analysis effort[18]. Lower scores were found in phonemic synthesis, phonemic manipulation and

phonemic transposition. This can be explained by the fact that phonemes are the smallest units of the language (which can also explain the difficulty to perceive them) and that phonetic and phonemic segments require a high degree of analytical capacity. Nevertheless, possible relations between phonemic awareness and syllabic skills in children with WS are still not well established[2]. The impaired phonological awareness skills found in the group with WS can be part of the typical language phenotype of the syndrome, in which there is often sophisticated domain of syntax and vocabulary, while metacognitive deficits are also present, once the individuals themselves do not understand the immediate implications of phrases and sentences they say[14,15,26].

Both age and level of education influence the development of phonological awareness, that is, these two factors contribute to the development of metaphonolofical competences[27]. However, Pearson correlation analysis of performance and variables such as age, level of education (1st to 8th grade) and IQ did not reveal significant correlations. The findings corroborate previous studies[28,29]. According to normative data[25] there is a progression in scores related to higher level of education in all PAT-OP subtests and that determines a growth in total correct answers. Individuals with WS, on the other hand, present irregularity in all analyzed items; this shows that, unlike the control group, for this group there is no relation between level of education and number of correct answers at PAT-OP.

Data from the present study was analyzed without considering the participants' ability to read and write. Phonological awareness depends on the writing system that is being taught and phonemic awareness appears to be achieved only with the introduction of an alphabetic system[18-21,25,27]. This capacity gradually develops as the child experiments ludic situations and is formally instructed in grapho-phonemic activities[17]. Thus, we can interpret that, when children become aware, analyze and manipulate segments or pieces of speech, besides having perceived that oral language is made of words, syllables and phonemes, they have developed or are developing and using their phonological awareness[18-21,25]. It is still possible to consider that the learning process is not the same and does not occur at the same time for all children, and this difference can be directly related to intrinsic factors, which, in their turn, depend on environmental, socioeconomic and cultural factors.

CONCLUSION

Data from this study corroborate the findings of literature that suggest that individuals with WS present difficulties in phonological processing tasks. These results have important implications for intervention programs and educational planning aimed at teaching writing and reading skills for children with WS. It is necessary, for instance, to consider in such programs the fact that this population presents intellectual disability, which is particularly important in Brazilian context where inclusion process present severe flaws for the education of children with special needs. It is also necessary to include continuous stimulations programs, with early interventions for children with WS in preschool age, adapted teaching methods and curriculum.

Further research is required to broaden the characterization of the profile of phonological processing skills in WS. Some aspects that could be contemplated in further studies are: larger sample; more detailed metalinguistic analysis; assessment of reading and writing skills for those who attend elementary school and effects of interventions using phonic method to develop phonological awareness skills.

RESUMO

Objetivos: avaliar o desempenho de crianças e adolescentes com Síndrome de Williams em tarefas de consciência fonológica e analisar esse desempenho em função da idade, escolaridade e indicadores de habilidade intelectual. **Métodos:** vinte e duas crianças e adolescentes (11 meninos e 11 meninas) com idades entre 7 e 18 anos realizaram os subtestes cubos e vocabulário das Escala Wechsler de Inteligência para estimativa de habilidades intelectuais e responderam a Prova de Consciência Fonológica por produção oral. **Resultados:** os participantes obtiveram valores do quociente de inteligência estimado compatível com rebaixamento intelectual. Nas habilidades de consciência fonológica os resultados mostraram que os participantes com a síndrome obtiveram desempenho rebaixado em nove dos dez subtestes da prova de consciência fonológica em relação à pontuação padronizada em função da idade, assim como do nível escolar. Não foram encontradas correlações significantes entre consciência fonológica, indicadores de habilidade intelectual, idade e escolaridade. **Conclusão:** os resultados corroboram os encontrados em pesquisas internacionais sugerindo rebaixamento em consciência fonológica na Síndrome de Williams. Dada a relevância desses achados é necessário incluir programas de estimulação contínua, inclusive com intervenções precoces dirigidas a crianças com Síndrome de Williams em idade pré-escolar, adequação de métodos de ensino e de currículo adaptado.

DESCRITORES: Síndrome de Williams; Deficiência Intelectual; Consciência

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