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Language Development in Rare Disease: Angelman Syndrome Vs Prader-Willi Syndrome

Esther Moraleda- Sepúlveda*, Sara Cañas-Pedrosa and Patricia López-Resa

Faculty of Health Science, University of Castilla-La Mancha, Spain

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***Corresponding author:** Esther Moraleda- Sepúlveda, Faculty of Health Science, University of Castilla-La Mancha, Avda Real Fábrica de la Seda s/n. 45600 Talavera de la Reina, Spain Email: esther.moraleda@uclm.es

Abstract

Angelman Syndrome (AS) and Prader-Willi Syndrome (PWS) are considered rare genetic disorders that share the same chromosomal region: 15q11.2-q13. This is why both share some common characteristics, such as, for example, delay in language development. However, there is still little research that specifically focuses on the linguistic profile in these populations. Therefore, the objective of this study was to know the characteristics of oral and written language that Angelman Syndrome and Prader-Willi Syndrome present from the point of view of parents. The sample consisted of 36 families (with children between 6 and 17 years old), of which 23 had children with AS and 13 had children with PWS. All of them answered the Language Assessment Scale of the standardized test CELF-4, Spanish Clinical Evaluation of Language Fundamentals-4 [1]. The scale is made up of 40 items that assess the perception of parents in areas such as: difficulty of listening, speaking, reading and writing. The results indicate that the majority of parents manifest problems in almost all the sub-areas related to oral language and written language, taking into account that many do not achieve a literacy level, with similar results in the comparison between both syndromes. These data support the importance of working on oral language delay and its relationship with the subsequent learning of literacy throughout its development.

Keywords: Angelman syndrome; Prader-willi syndrome; Development; Language

Introduction

Uncommon diseases owe their name to the low prevalence of their occurrence in the population, and European legislation considers a disease to be "rare" when it is less than 1:2000 [2,3]. Although it is difficult to pinpoint an exact figure, approximately 6-8% of the world's population has a rare disease [4].

The Angelman and Prader-Willi syndromes (hereinafter AS and PWS) are two genetic diseases that are considered uncommon. Both syndromes are caused by the lack of expression of the chromosomal region's imprinted genes 15q11-q13. In the case of AS, the loss of information takes place in the maternal chromosome while in PWS the silencing occurs in the chromosome coming from the father [5-7].

Broadly speaking, AS is characterized by severe mental disability, developmental delay, ataxia, microcephaly and frequent smiling and laughing [8,9]. Physically, people with AS tend to have blue eyes and skin hypopigmentation, low waist, microcephaly, and macroglossia [10,11]. Furthermore, around 90% of this population develops epileptic seizures during their lifetime [12]. The linguistic level of people with AS is characterized by specific difficulties in language development, especially at the expressive level (written and verbal). Most have minimal or no speech development, limited to two or three words [11,13]. However, some research has reported that certain people with AS are able to functionally use augmentative and alternative communication (AAC) systems [14,15]. In addition, although natural gestures are not strictly considered to be an AAC system, they have been shown to be effective in supporting language development and, specifically, the subsequent implementation of an AAC system in this population [16].

For their part, people with PWS possess a phenotypic profile with characteristic facial features, short stature, neonatal hypotonia, hypogonadism, hyperphagia, obesity and deficits at the visual and bone levels, often exhibiting scoliosis [17,18]. Cognitive-behaviorally, they manifest motor and language delay, intellectual disability to some degree, and behavioral problems that include stubbornness, anger, and tantrums [19,20]. At the linguistic level, language development is often delayed, especially at the verbal level. These difficulties include deficits in articulation, phonological sequencing, and writing [21]. Some

studies have also pointed to a delayed start of the syntax that progresses favorably as development progresses [22,23]. On the other hand, the decoding processes are favorable, as well as reading comprehension.

The scarce scientific literature highlights the differences in their linguistic symptoms, although there is a patent need to delve into their language profiles. Therefore, the goal of our study was to explore the characteristics of the oral and written language of people with AS and PWS.

Method

Participants

The sample consisted of a group of 36 families of children and adolescents aged 6 to 17 years, of which 23 had a son or daughter diagnosed with Angelman syndrome (AS) (12 women and 11 men) whose average age was 12.0 years, and 13 had a son or daughter with Prader-Willi syndrome (PWS) (6 women and 7 men) whose average age was 11.0 years.

The family members of the children with AS were mostly the children's mothers, with only one father participating. Regarding the family members of the people with PWS, all were mothers except three fathers.

For children with AS, the accredited percentage of disability was between 37% and 100% and for children with PWS, the percentage ranged between 10% and 66%.

Instrument

The Language Assessment Scale of the "CELF-4, Spanish Clinical Evaluation of Language fundamentals-4" standardized test [1] was used to carry out this study.

This is a test aimed at the population aged between 5 and 21 years that assesses listening, speaking, reading and writing using a Likert-type scale. The scale is made up of 40 items that assess the perception of parents, caregivers and teachers.

Process

First, multiple AS and PWS associations were contacted and informed about the study, receiving detailed information on the project. In order to obtain greater participation, the questionnaire was conducted virtually and sent in electronic format through a link that redirected participants to the online form.

The research was carried out with the express consent of the participants. It is worth mentioning that this study complies with the provisions contained in the Declaration of Helsinki on ethical principles for medical research involving human subjects.

Results

The data obtained were analyzed taking into account the four areas into which the questionnaire is divided and the two groups of participants.

Listening

Table 1: Listening-related difficulties.

		1	2	3	4	US*
Trouble paying attention	AS	0%	26.09%	56.52%	17.39%	0%
	PWS	7.90%	46.15%	23.98%	23.08%	0%
Trouble following oral instructions	AS	8.70%	63.87%	21.74%	4.25%	0%
	PWS	15.83%	69. 23%	7. 69%	7.69%	0%
Trouble remembering what is said	AS	17.39%	47.83%	21.74%	8.70%	4.35%
	PWS	30.77%	23.08%	46.15%	0%	0%
Trouble understanding what is said	AS	34.78%	60.87%	4.35%	0%	0%
	PWS	46. 15%	38. 46%	7.69%	0%	0%
Needs to ask for what has been said to be repeated	AS	30.43%	21.74%	13.04%	0%	34.78%
	PWS	30.77%	46.15%	15.38%	7.69%	0%
Trouble understanding the meanings of words	AS	8.70%	52.17%	17.39%	8.70%	13.04%
	PWS	38.46%	46.15%	7.69%	7.68%	0%
Trouble understanding new concepts	AS	0%	43.48%	21.74%	21.74%	13.04%
	PWS	23.08%	23.08%	38.46%	15.38%	0%
Trouble looking at others when speaking or listening	AS	52.17%	34.78%	4.35%	0%	8.70%
	PWS	23.08%	46.15%	15.38%	15.38%	0%
Trouble understanding facial expressions, gestures, or body language	AS	56.52%	34.78%	0%	0%	8.70%
	PWS	53.85%	15.38%	23.08%	7.69%	0%
*Undeveloped skill						

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With regard to understanding, although people with AS and PWS seem to exhibit difficulties in all listening-related items, the results show how people with AS develop fewer skills than people with PWS. The results indicate that people with AS have attentional and verbal language comprehension difficulties. In this line, it was possible to observe how, from the point of view of the parents, people with AS have not acquired certain skills such as understanding the meanings of words and new concepts, looking at others when they are spoken or listened to and understanding facial expressions, gestures and body language.

For their part, people with PWS experience particular difficulties in following oral instructions, in understanding what is being said to them, and in understanding new concepts. Table 1 reflects in percentage terms the response frequency of both groups.

Speaking

Concerning language expression, the results show how, despite the fact that people with AS and PWS exhibit difficulties in this area, these are more pronounced in the case of AS.

In this line, the data have revealed that more than 30% of the participants with AS had not developed 12 of the 19 skills that were asked about. In addition, those participants who reported having developed them reported having difficulties often or always or almost always.

Table 2: Speech-related difficulties.

		1	2	3	4	US*
Trouble answering what is asked	AS	0%	30.43%	34.78%	26.09%	8.70%
	PWS	46.15%	38.46%	7.69%	7.69%	0%
Trouble answering questions as quickly as other students	AS	0%	4.35%	21.74%	65.22%	8.70%
	PWS	15.38%	23.08%	46.15%	15.38%	0%
Trouble asking for help when needed	AS	26.09%	39.13%	21.74%	13.04%	0%
	PWS	46.15%	23.08%	23.08%	7.69%	0%
Trouble asking questions	AS	4.31%	0%	13.04%	56.52%	26.09%
	PWS	61.54%	23.08%	7.69%	7.69%	0%
Trouble using varied vocabulary when speaking	AS	0%	4.35%	17.39%	43.48%	34.78%
	PWS	53.85%	15.38%	15.38%	15.38%	0%
Trouble thinking of (finding) the right word	AS	0%	0%	17.39%	56.52%	26.09%
	PWS	15.38%	53.85%	23.08%	7.69%	0%
Trouble expressing thoughts	AS	4.35%	8.70%	21.74%	34.78%	30.43%
	PWS	38.43%	15.38%	15.38%	33.77%	0%
Trouble describing things to others	AS	0%	4.35%	13.04%	47.83%	34.78%
	PWS	23.08%	53.85%	7.69%	15.38%	0%
Trouble focusing on the topic of conversation	AS	4.35%	30.43%	17.39%	26.09%	21.74%
	PWS	30.77%	46.15%	0%	23.08%	0%
Trouble focusing on the most important aspects when speaking	AS	4.35%	13.04%	4.35%	39.13%	39.13%
	PWS	7.69%	53.85%	15.38%	23.08%	0%
Trouble ordering events properly when telling a story or talking about an event	AS	0%	8.70%	4.35%	47.48%	39.13%
	PWS	7.69%	8.46%	30.77%	3.08%	0%
Uses poor grammar when speaking	AS	0%	4.31%	4.31%	60.87%	30.43%
	PWS	56.85%	23.08%	15.38%	7.69%	0%
Trouble forming complete sentences when speaking	AS	0%	4.31%	4.31%	52.17%	39.13%
	PWS	61.54%	7.69%	23.08%	7.69%	0%
Speaks in short sentences, without grammatical ties	AS	17.39%	0%	13.04%	34.78%	34.78%
	PWS	56.85%	15.38%	23.08%	7.69%	0%
Trouble expanding on an answer or giving details when speaking	AS	0%	4.31%	8.70%	47.83%	39.13%
	PWS	46.15%	15.38%	23.08%	15.38%	0%

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Trouble having a conversation with someone	AS	0%	8.70%	4.31%	52.17%	34.78%
	PWS	69.23%	7.69%	15.38%	7.69%	0%
Trouble talking to a group of people	AS	0%	4.31%	8.70%	47.83%	39.13%
	PWS	61.54%	7.69%	15.38%	15.38%	0%
Trouble saying something in a different way when someone does not understand	AS	0%	8.70%	13.04%	43.48%	34.78%
	PWS	23.08%	56.85%	7.69%	15.38%	0%
Gets upset when not understood	AS	8.70%	17.39%	30.43%	34.78%	8.70%
	PWS	7.69%	46.15%	38.46%	7.69%	0%
*Undeveloped skill						

Regarding the participants with PWS, the results show more problems when answering questions as quickly as other children, difficulties in accessing vocabulary and when expressing their feelings. Along these lines, the data offer us a fairly broad perspective on the lexical-semantic and morphosyntactic difficulties that people with PWS exhibit. Unlike participants with AS, all participants with PWS had developed all of the speech-related skills. Table 2 reflects the frequency of responses to each speech-related item in terms of frequency.

Reading

Next, concerning reading, the results again demonstrated how a high percentage of the participants with AS have not acquired reading-related skills.

Regarding the participants with PWS, the results demonstrated how they exhibit particular difficulties in explaining what they have read and following written instructions. Table 3 reflects in percentage terms the frequency of responses referring to the reading section.

Table 3: Reading-related difficulties.

		1	2	3	4	US*
Trouble pronouncing words when reading	AS	0%	0%	0%	56.52%	43.48%
	PWS	38.46%	30.77%	23.08%	7.69%	0%
Trouble understanding what is being read	AS	0%	4.31%	4.31%	47.83%	43.48%
	PWS	30.77%	46.15%	15.38%	7.69%	0%
Trouble explaining what is being read	AS	0%	0%	4.31%	47.83%	47.83%
	PWS	7.69%	46.15%	38.46%	7.69%	0%
Trouble identifying what is being read	AS	0%	13.04%	0%	43.48%	43.48%
	PWS	33.77%	38.46%	23.08%	7.69%	0%
Trouble identifying the main idea	AS	0%	4.31%	0%	52.17%	43.48%
	PWS	23.08%	30.77%	23.08%	23.08%	0%
Trouble remembering details	AS	0%	4.31%	8.70%	43.48%	43.48%
	PWS	0%	23.08%	30.77%	23.08%	0%
Trouble following written instructions	AS	0%	4.31%	0%	52.17%	43.48%
	PWS	23.08%	46.15%	30.77%	0%	0%
*Undeveloped skill						

Writing

To conclude, concerning writing, data have proved that the severity of the difficulties is considerably greater in participants with AS. In this line, it has been seen that approximately half of the participants with AS do not develop writing skills and those who do exhibit very significant difficulties in carrying out activities involving writing. Hence, the category "always or almost always" presents a high response rate.

In contrast, 100% of the participants with PWS reported having developed writing skills, although deficits were observed in all the skills that were asked after, especially when it came to ordering words correctly when writing sentences and using poor grammar.

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Table 4: Writing-related difficulties.

		1	2	3	4	US*
Trouble writing their thoughts	AS	0%	0%	0%	52.17%	47.83%
	PWS	23.08%	15.38%	30.77%	30.77%	0%
Poor grammar when writing	AS	0%	0%	0%	52.17%	47.83%
	PWS	30.77%	15.38%	23.08%	30.77%	0%
Trouble writing complete sentences	AS	0%	0%	0%	52.17%	47.83%
	PWS	46.15%	15.38%	7.69%	30.77%	0%
Writes in short sentences, without grammatical ties	AS	8.70%	0%	0%	43.48%	47.83%
	PWS	38.46%	26.08%	15.38%	23.08%	0%
Trouble expanding an answers or giving details when writing	AS	0%	0%	0%	52.17%	47.83%
	PWS	15.38%	30.77%	23.08%	30.77%	0%
Trouble ordering words correctly when writing sentences.	AS	0%	0%	0%	52.17%	47.83%
	PWS	38.46%	23.08%	7.69%	30.77%	0%
*Undeveloped skill						

Discussion

Although other authors have recently conducted studies comparing the linguistic profile of AS and PWS [24,25], there are very few studies exclusively focused on comparing the oral and written language of the population with Angelman syndrome and Prader-Willi syndrome. Thus, the results allow us to observe how, although people with both syndromes experience difficulties in oral and written language, these are more severe in the case of AS.

Concerning AS, it was observed that most of the participants with this syndrome had not managed to develop oral language, and also had reading and writing deficits. However, the data shows how these people are able to communicate to ask for help. This has been observed in other research conducted with this population, such as the one carried out by Perrino et al. [26], who noted that people with AS exercise the basic pragmatic function of asking for help by banging on surfaces or calling out. The results of this study follow the line of other research regarding the development of oral language. For example, Grieco et al. [27] observed that people with AS obtain a basic percentile in language when the Bailey scale is administered. For their part, Quinn et al. [28] determined that people with AS present a non-verbal communication level that is much higher than its verbal counterpart. Lastly, Perrino et al. [26] reported that children with AS have little verbal initiative and few vocalizations.

These communication deficits are also consistent with a study by Adams et al. [29] in which they wanted to monitor the perceptions of parents of children with AS of their children's treatments. In this case, the results showed that almost half of them considered communication skills to be a priority issue, which implies that, as was observed in this study, these skills are highly afflicted, and therefore this is a common concern among family members.

Concerning PWS, the data suggest the existence of short-term memory difficulties, as well as attentional difficulties. Likewise, the results demonstrated the existence of phonetic-phonological and articulatory difficulties, in line with previous studies [30]. In this line, Dimitropoulos et al. [31] recorded a better performance in tasks measuring comprehensive versus expressive language.

Despite the fact that both syndromes exhibit oral and written linguistic difficulties, the data have shown how the difficulties are more severe in AS, although it would be convenient to analyze, in subsequent studies, how these difficulties evolve.

Therefore, we believe it would be interesting to continue researching the linguistic profile of these two syndromes, making it possible to implement speech therapy intervention programs that allow oral and written language skills to be improved, producing a direct impact on the quality of life of people with AS and PWS.

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