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Speech-language Pathology Characteristics of Patients Assisted by a Clinical Genetics Service

Características Fonoaudiológicas de Pacientes Atendidos por um Serviço de Genética Clínica

Características de patología del habla y lenguaje de Pacientes Atendidos por un Servicio de Genética Clínica

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Abstract

Introduction: The collaborative efforts of medical genetics and speech therapy are essential, contributing to the development of procedures that assist in treating patients with communication disorders. **Objective:** To analyze the speech therapy characteristics of pediatric patients seen by a clinical genetics service. **Methods:** Observational cross-sectional study conducted with patients seen at the genetics service of a hospital in Porto Alegre. A questionnaire related to hearing, swallowing, orofacial motricity, voice, and language areas was used for data collection. **Results:** The sample consisted of 54 participants aged between 8 months and 17 years, with an average age of 6 years and 5 months. 24.07% (n=13) of the patients had a diagnosis of syndrome, and 59.26% (n=32) had delayed neuropsychomotor development. Regarding the speech therapy profile, 81.48% (n=44) had some harmful oral habit during childhood. 16.67% (n=9) reported some difficulty in hearing, and 29.62% (n=16) in swallowing. 85.19% (n=46) of the participants showed developed oral language, and of these, 71.74% (n=33) made speech substitutions. 33.33% (n=18) of the patients were already undergoing speech therapy, and another 24.07% (n=13) were on the waiting list for this treatment. **Conclusions:** A significant portion of the patients presented

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Authors' contributions:
ICS: data collection; study outline;
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complaints and/or manifestations in the areas of human communication, especially regarding language, speech, and harmful oral habits. These data highlight the importance of referral to the speech therapy team.

Keywords: Medical Genetics; Syndrome; Speech Therapy; Language Development Disorders

Resumo

Introdução: O trabalho conjunto da genética médica e da fonoaudiologia é essencial, contribuindo para o desenvolvimento de procedimentos que auxiliam no tratamento de pacientes com distúrbios da comunicação. **Objetivo:** Analisar as características fonoaudiológicas de pacientes pediátricos atendidos por um serviço de genética clínica. **Método:** Estudo transversal observacional, realizado com pacientes atendidos pelo serviço de genética de um hospital em Porto Alegre. Para a coleta de dados, aplicou-se um questionário relacionado as áreas de audição, deglutição, motricidade orofacial, voz e linguagem. **Resultados:** A amostra foi constituída por 54 participantes com idades entre 8 meses e 17 anos (média de idade 6 anos e 5 meses). 24,07% (n=13) dos pacientes apresentaram diagnóstico de síndrome, e 59,26% (n=32) tinham atraso no desenvolvimento neuropsicomotor. Com relação ao perfil fonoaudiológico, 81,48% (n=44) apresentaram algum hábito oral deletério durante a infância. 16,67% (n=9) percebiam alguma dificuldade para ouvir e 29,62% (n=16) para deglutir. 85,19% (n=46) dos participantes manifestaram a linguagem oral desenvolvida e, destes, 71,74% (n=33) apresentavam trocas na fala. 33,33% (n=18) já estavam em atendimento fonoaudiológico, e outros 24,07% (n=13) estavam na fila de espera para este atendimento. **Conclusões:** Uma parte significativa dos pacientes apresentou queixas e/ou manifestações nas áreas da comunicação humana, principalmente em relação à linguagem, à fala e aos hábitos orais deletérios. Esses dados destacam a importância do encaminhamento para a equipe de fonoaudiologia.

Palavras-chave: Genética Médica; Síndrome; Fonoaudiologia; Transtornos do Desenvolvimento da Linguagem

Resumen

Introducción: La colaboración entre genética médica y foniatría es esencial para desarrollar procedimientos que ayuden en el tratamiento de pacientes con trastornos de la comunicación. **Objetivo:** Analizar las características de patología del habla y lenguaje de pacientes pediátricos atendidos por un servicio de genética clínica. **Método:** Estudio transversal observacional con pacientes atendidos por el servicio de genética de un hospital en Porto Alegre. Se aplicó un cuestionario sobre audición, deglución, motricidad orofacial, voz y lenguaje. **Resultados:** La muestra consistió en 54 participantes con edades comprendidas entre 8 meses y 17 años (media: 6 años y 5 meses). El 24,07% (n=13) de los pacientes tenían un diagnóstico de síndrome, y el 59,26% (n=32) presentaron retraso en el desarrollo neuropsicomotor. En cuanto al perfil foniatra, el 81,48% (n=44) presentaron algún hábito oral perjudicial durante la infancia. El 16,67% (n=9) reportaron dificultades para oír, y el 29,62% (n=16) para tragar. El 85,19% (n=46) manifestaron lenguaje oral desarrollado y, de ellos, el 71,74% (n=33) realizaban intercambios en el habla. El 33,33% (n=18) de los pacientes ya estaban en tratamiento foniatra y el 24,07% (n=13) estaban en lista de espera para este tratamiento. **Conclusiones:** Una parte significativa de los pacientes presentó quejas y/o manifestaciones en las áreas de la comunicación humana, especialmente en relación con el lenguaje, el habla y los hábitos orales perjudiciales, enfatizando la importancia de la derivación al equipo de foniatría.

Palabras clave: Genética Médica; Síndrome; Foniatría; Transtornos del Desarrollo del Lenguaje

Introduction

Medical genetics focuses on individually rare conditions that represent a significant group of diseases with increasingly relevant implications for community health[1]. In this sense, advances in genetics in recent decades have provided a better understanding of various diseases, enhancing the understanding of their causes and effects, improving diagnostic methods, and developing innovative therapeutic practices[2,3]. Considerable efforts are being made to detect genetically caused diseases early, allowing for prenatal or postnatal diagnoses[2].

In this context, Speech-Language Pathology emerges as one of the essential specialties, being the science responsible for studying human communication and its disorders[4]. The speech-language pathologist plays a crucial role in promoting health, preventing, evaluating, and diagnosing, as well as guiding, providing therapy (habilitation and rehabilitation), and enhancing aspects related to oral and written language, voice, hearing, orofacial motricity, and swallowing[5,6]. Genetic syndromes can affect various aspects of physical, cognitive, and linguistic development, including communication. Communication disorders caused by genetic syndromes can have a significant impact on an individual's social life, including language impairment, speech issues, oral functions, and swallowing difficulties[7]. Therefore, the role of the speech-language pathologist is fundamental in the early diagnosis and treatment of genetic disorders, in order to provide interventions that improve communication, socialization, and the quality of life of affected patients.

An example of this interface is Down syndrome, or Trisomy of chromosome 21, which is one of the most studied and frequent chromosomal abnormalities in humans[8]. Among various characteristics, individuals with this syndrome may also present implications in language, speech, voice, sequential auditory memory, and sometimes hearing loss, bringing several problems related to their socialization[7,9]. Studies show that appropriate speech-language intervention in the first months of life can favor the development of receptive and expressive language, the oral sensorimotor system, and stomatognathic functions, contributing to the overall development, providing greater independence within their limitations, and improving their social performance[9].

Moreover, there is still no effective treatment available for a large part of genetic diseases. Only 10% of them have specific medicinal options, many of which are high-cost[10]. Additionally, these diseases are characterized by a diversity of signs and symptoms that vary not only among different diseases but also among patients affected by the same disease, which hinders diagnosis and future intervention[11]. Treatment often requires multidisciplinary follow-up, involving medical geneticists, physiotherapists, speech-language pathologists, nutritionists, and psychologists, among others, aiming to alleviate symptoms or delay their onset[11].

Therefore, this study aimed to analyze the speech-language characteristics of pediatric patients seen in a clinical genetics service.

Material and Methods

This is a cross-sectional observational study conducted with pediatric patients attended by the Clinical Genetics Service at Hospital da Criança Santo Antonio (HCSA), em Porto Alegre, Rio Grande do Sul. This study was approved by the Research Ethics Committee of Irmandade Santa Casa de Misericórdia de Porto Alegre (under the Decision No. 5.339.364) and registered with ComPesq *[Research Committee]* (No. 62/2022). Data collection was carried out from July 2022 to June 2023.

At first, all patients attended by the Genetics Service at HCSA ambulatory, belonging to the Unified Health System (SUS), were consecutively invited to participate in this study after the presentation of the objectives and data collection methods. All information had to be completed and the patients' guardians had to sign the Informed Consent Form (ICF) in order to be included in this study.

Data were collected through a questionnaire administered by speech-language pathology students trained for this purpose. The questionnaire applied was specifically designed for this research based on the literature[12,13]. The questionnaire consisted of four parts: patient history (identification data), health problems, neuropsychomotor development data, and questions related to the perception of possible speech-language pathologic alterations. The questionnaire was developed to conduct an initial screening and contained questions related to the areas of hearing, voice, swallowing, orofacial

motricity/deleterious oral habits, and language/speech.

The first part of the questionnaire addressed topics such as the patient's name, age, sex, syndrome diagnosis, complaint/referral, gestational age, Apgar score, pregnancy and/or birth complications, presence of malformations, use of medications, and whether the patient attends a rehabilitation center or receives treatment from other professionals. Next, health problems were recorded, divided into neurological, orthopedic, digestive, hormonal, and other categories. Regarding neuropsychomotor development data, responses were categorized as normal or altered, taking into account the corresponding ages for cervical control, sitting without support, crawling, and walking, based on medical evaluation.

With respect to the questions related to the perception of speech-language pathologic alterations, this stage was divided into five parts. First, questions related to hearing were asked, such as the perception of difficulty in hearing, presence of tinnitus and/or dizziness, history of otitis, and the performance of hearing tests and their results. The second part included questions about voice, addressing the occurrence of voice loss and hoarseness.

For the third part, questions about swallowing were asked, investigating possible difficulties, presence of cough, choking and/or residues after swallowing, gastroesophageal reflux, and other problems. Subsequently, aspects related to breastfeeding were addressed, such as the history or possibility of breastfeeding and the use of a bottle, as well as questioning how long the child practiced these habits.

The fourth part of the questionnaire covered deleterious oral habits, including the use of a pacifier, finger sucking, bruxism, biting the oral mucosa, and biting objects. The age until which the patient practiced these habits was asked. Finally, the fifth part included questions related to language and speech, investigating difficulties in attention/concentration, memory, and interaction, as well as schooling, language intelligibility, delays, and speech exchanges.

After data collection, the results were tabulated and stored in a spreadsheet using Microsoft Office Excel® v.16.75. Subsequently, data was statistically analyzed regarding absolute frequency, mean, and median.

Results

During the study period, 59 patients were attended at the Medical Genetics Service of HCSA. Of these, 5 were excluded due to a lack of information or absence of a signed ICF. Thus, 54 patients were analyzed and included in the present study. Among this group, 51.85% (n=28) were male, and the patients' ages ranged from 8 months to 17 years, with an average of 6 years and 5 months (median of 6 years and 2 months). Patients from 40 different municipalities were included in the study, with a predominance of patients from the city of Porto Alegre (n=10).

Regarding syndrome diagnosis, 24.07% (n=13) of the patients had a confirmed diagnosis through tests, and a wide variety of diseases were found. (TABLE 1)

Table 1. Characterization of the sample by syndrome diagnosis (n=54) and description of identified pathologies (n=13)

Variable	N	%
Diagnosis		
Yes	13	24.07
No	41	75.93
Pathology		
Dystrophinopathy	1	7.69
Goldenhar Syndrome	1	7.69
Down Syndrome	1	7.69
Angelman Syndrome	1	7.69
Peutz-Jeghers Syndrome	1	7.69
Tuberous Sclerosis	1	7.69
Distal Arthrogryposis	1	7.69
16p11.2 Microduplication	1	7.69
Rett Syndrome	1	7.69
Apert Syndrome	1	7.69
Albinism	1	7.69
Alagille Syndrome	1	7.69
Cellular Mesoblastic Nephroma	1	7.69

Of those without a confirmed genetic diagnosis (n=41), 24.39% (n=10) had a possible diagnosis, with Autism Spectrum Disorder being the most common, accounting for 7.32% (n=3) of the cases. The remaining cases (n=7) presented various hypotheses: Ehlers-Danlos Syndrome (n=1), Silver-Russell Syndrome (n=1), Fragile X Syndrome (n=1), Inborn Error of Metabolism (n=1), Epilepsy (n=1), Goldenhar Syndrome (n=1), and Neurofibromatosis (n=1).

Additionally, it was observed that 37.04% (n=20) of the patients had at least one congenital malformation. Most common malformations were congenital clubfoot and cardiopathy, each with a 7.41% (n=4) occurrence. (TABLE 2)

Table 2. Identification of congenital malformations* (n=54)

Variable	N	%
Presence of malformation		
Yes	20	37.04
No	34	62.96
Malformation		
Congenital clubfoot	4	7.41
Congenital heart disease	4	7.41
Cleft lip and palate	2	3.70
Spina bifida	2	3.70
Anophthalmia	1	1.85
Gastroschisis	1	1.85
Distal arthrogryposis	1	1.85
Congenital scoliosis	1	1.85
Tracheal stenosis	1	1.85
Hemifacial microsomia	1	1.85
Unilateral type 3 microtia	1	1.85
Craniosynostosis	1	1.85
Dandy-Walker syndrome	1	1.85
Complete syndactyly of hands	1	1.85
Complete syndactyly of feet	1	1.85
Multicystic kidney	1	1.85
Ambiguous genitalia	1	1.85
Grade 4 hypospadias	1	1.85
Congenital ptosis	1	1.85
Chiari malformation type II	1	1.85
Congenital mesoblastic nephroma	1	1.85

*Patients could had more than one malformation.

Regarding neuropsychomotor development, 59.26% (n=32) of the patients presented some delay, according to medical evaluation. Moreover, in terms of health problems, 50% (n=27) had neurological problems, 29.63% (n=16) had orthopedic problems, 18.52% (n=10) had visual problems, and 33.33% (n=18) had some other deficiency.

Regarding speech-language pathology alterations, specifically hearing, all guardians of the patients were questioned about their perceptions related to the patients' hearing, with 16.67% (n=9) perceiving some difficulty in hearing. (TABLE 3)

Table 3. Audiological aspects identified in the interview (n=54)

Variable	N	%
Perceived difficulty hearing		
Yes	5	9.26
Sometimes	4	7.41
No	45	83.33
Presence of tinnitus		
Yes	0	0
Sometimes	0	0
No	31	57.41
Not applicable	23	42.59
Presence of dizziness		
Yes	1	1.85
Sometimes	3	5.56
No	29	53.70
Not applicable	21	38.89
History of otitis		
Yes	3	5.56
Sometimes	5	9.26
No	46	85.18
Underwent hearing test		
Yes	45	83.33
No	9	16.67

Furthermore, the hearing tests performed by each patient were analyzed. It was found that 83.33% (n=45) of the patients had already undergone some hearing test. (TABLE 4)

Table 4. Results of analyzed hearing tests (n=45)

Hearing tests (n=45)	Number of cases	Regular	Changed	No information
Baby hearing screening	45	35 (77.78%)	6 (13.33%)	4 (8.89%)
Audiometry	8	4 (50%)	3 (37.5%)	1 (12.5%)
BAEP*	8	3 (37.5%)	3. (37.5%)	2 (25%)
CAP**	2	0	2 (100%)	0

* Brainstem Auditory Evoked Potential
** Evaluation of central auditory processing
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Concerning voice-related aspects, 24.07% (n=13) of the patients reported vocal alterations only in the presence of respiratory diseases, such as colds and flu. Regarding swallowing and feeding, 29.62% (n=16) of the patients perceived some difficulty in swallowing, with the most common characteristic being the presence of cough after swallowing. (TABLE 5)

Table 5. Aspects related to swallowing function identified in the interview (n=54)

Variable	N	%
Difficulty swallowing		
Yes	7	12.96
Sometimes	9	16.67
No	38	70.37
Cough after swallowing		
Yes	9	16.67
Sometimes	7	12.96
No	38	70.37
Choking during meals		
Yes	6	11.11
Sometimes	6	11.11
No	42	77.78
Gastroesophageal Reflux		
Yes	8	14.81
Sometimes	4	7.41
No	42	77.78
Residues after swallowing		
Yes	5	9.26
Sometimes	3	5.56
No	46	85.18

Additionally, regarding breastfeeding, 61.11% (n=33) were breastfed, having stopped the practice, on average, at 14 months. As for bottle-feeding, 77.78% (n=42) used a bottle until, on average, 3 years and 5 months of age. Concerning deleterious oral habits, 81.48% (n=44) reported having maintained some habit during childhood. The most common was pacifier use, with 50% (n=27). (TABLE 6)

Table 6. Deleterious oral habits identified in the interview (n=54)

Variable	N	%	Mean age
Presented any oral habit			
Yes	44	81.48%	
Pacifier use			
Yes	27	50%	
If yes, until when			2y9m
Thumbsucking			
Yes	8	14.81%	
If yes, until when			4y8m
Bruxism			
Yes	24	44.44%	
If yes, until when			5y4m
Biting oral mucosa			
Yes	6	11.11%	
If yes, until when			5y7m
Biting objects			
Yes	15	27.78%	
If yes, until when			3y5m

*y = years; m = months.

In the area of language, 85.19% (n=46) of the patients interviewed had developed oral language. Among the 14.81% (n=8) who did not have oral language, 5.56% (n=3) were under 18 months old; 3.70% (n=2) were between 2 and 3 years old; and 3.70% (n=2) were over 13 years old. Additionally, the 46 participants with oral language were questioned about speech intelligibility, sentence formation, storytelling, and speech exchanges. (TABLE 7)

Tabela 7. Aspectos relacionados à linguagem oral identificados na entrevista (n=46)

Variable	N							
	0 - 1y*	1y1m - 2y*	2y1m - 3y*	3y1m - 5y*	5y1m - 6y*	6y1m - 10y*	10y1m - 17y*	Total
Everyone understands the child's speech								
Yes	0	0	0	1	7	6	14 (30.44%)	
Sometimes	0	0	1	1	0	1	0	3 (6.52%)
No	0	9	5	3	0	7	5	29 (63.04%)
Forms sentences								
Yes	0	0	1	1	1	10	9	22 (47.83%)
Sometimes	0	0	2	2	0	1	0	5 (10.87%)
No	0	9	3	1	0	4	2	19 (41.30%)
Tells stories								
Yes	0	0	1	0	1	10	8	20 (43.48%)
Sometimes	0	0	0	2	0	0	0	2 (4.35%)
No	0	9	5	2	0	5	3	24 (52.17%)
Makes speech substitutions								
Yes	0	9	6	4	0	8	6	33 (71.74%)
Sometimes	0	0	0	0	0	2	2	4 (8.70%)
No	0	0	0	0	1	5	3	9 (19.56%)

*y = years; m = months.

Moreover, all participants were questioned regarding attention/concentration deficits, memory difficulties, and interaction difficulties. Thus, 44.44% (n=24) had attention/concentration deficits; 25.93% (n=14) had memory difficulties; and 33.33% (n=18) had interaction difficulties.

Finally, 88.89% (n=48) of the patients attended a rehabilitation center and/or received treatment from other health professionals. Among these, 37.07% (n=20) were under the care of a neurologist, 33.33% (n=18) were receiving speech therapy, and 22.22% (n=12) were being assisted by the physiotherapy team. In the specific context of speech-language pathology, 24.07% (n=13) of the patients were on the waiting list, while 5.56% (n=3) had already completed speech-language pathology sessions. This resulted in a total of 62.96% (n=34) of patients who had attended or were awaiting speech-language interventions.

Discussion

This study identified various characteristics and communication disorders in a sample of pediatric patients with suspected genetic syndromes. Despite this, a significant portion of these patients was receiving rehabilitation care, suggesting an adequate response to their needs. It should be noted that a limitation of this study is the small sample size, which, combined with the lack of confirmed syndrome or genetic condition diagnoses, prevented the description of specific speech-language alterations for each syndrome evaluated by the Clinical Genetics team. Additionally, the inclusion of control groups would allow for more precise comparisons.

The broad age range of the patients, from infants to adolescents, underscores the importance of genetic investigation at all ages, providing crucial insights for the diagnosis and management of genetic conditions. The broad geographical reach

of the sample, consisting of patients from 40 different municipalities, highlights the centralization of Medical Genetics services in major urban centers, which is an issue also found in other studies on genetics care in Brazil[14].

In addition, the low diagnosis rate of syndromes underscores the ongoing need for efforts to identify underlying causes. Diagnostic difficulties in genetic diseases are exacerbated by the similarity of signs and symptoms to common diseases and the complexity of genetic conditions, posing challenges for patients and their families in seeking diagnoses[11]. This complexity often results in significant delays for children to be referred to genetics services, further prolonging the onset of necessary investigations and interventions.

The prevalence of neuropsychomotor developmental delays, coupled with the diversity of health problems, emphasizes the multifaceted nature of genetic conditions, requiring comprehensive care approaches. Thus, it highlights the need for early and multidisciplinary interventions as well as continuous support for patients and their families. Studies reported that early stimulation has positive outcomes in cases of neuropsychomotor developmental delays, potentially reducing morbidity and mortality and enabling children to achieve their best possible development, while minimizing neuropsychomotor sequelae[14,15,16].

Regarding hearing, a considerable portion of the patients had already undergone some hearing test, notably the newborn hearing screening. According to various epidemiological studies, the prevalence of hearing impairment ranges from one to six newborns per thousand live births in Brazil, reaching four per hundred newborns from Neonatal Intensive Care Units[17]. Since 2010, with the publication of Federal Law No. 12.303, the Universal Newborn Hearing Screening (UNHS) is mandatory for all live newborns nationwide[18]. Therefore, the proper implementation of the newborn hearing test enables early detection of hearing pathologies, minimizing future difficulties in oral language development, maximizing linguistic and communicative competencies, and supporting literacy development[19]. However, the full realization of this initiative faces challenges related to access and the availability of materials and/or specialized professionals, potentially hindering its implementation for all live births. This consideration might explain the small portion of patients in our sample without recorded newborn hearing screening.

Additionally, 16.67% of caregivers reported perceiving some difficulty in the child's hearing. Studies have confirmed that parents are often the first ones to suspect hearing impairment in their children, making them a crucial factor in the screening of the pediatric population as a whole[20]. The initial behavioral symptoms manifested in children with hearing loss include irritability, reduced responses, inattention, and sleep disturbances[20,21]. Therefore, it is important to always consider parents' concerns about suspected hearing loss until this loss is convincingly ruled out or the reason for the child's different behavior is discovered[20].

Concerning deleterious oral habits, studies conducted with children aged four months to 13 years indicate a prevalence of 30.8% to 70.8%, with pacifier sucking being the most frequent habit[22,23], as also observed in the present study. Furthermore, it is recommended that habits be discontinued preferably before two years of age, as inadequacies can be identified after this period, potentially affecting both orofacial muscle function and dental occlusion, especially anterior open bite[23]. In this study, all analyzed habits persisted beyond this age range, indicating the potential need for interventions to correct and restore functions.

Regarding language, each age group is expected to present certain linguistic characteristics, based on the child's auditory development and exposure to the structure of the native language[24]. According to the literature, the child begins to say their first meaningful words from 12 to 18 months; then, from two to three years, the child should start forming sentences; and from four to five years, the child should be able to tell simple stories[25]. Thus, in this study, it was observed that a portion of the patients had delays in these aspects.

Research indicates that the most frequent demands for speech-language intervention in children are related to speech problems, especially phonological exchanges[19,26,27]. This finding is in line with the data from this study, as it was the most common complaint among the analyzed patients. Furthermore, the child is expected to pronounce words correctly from five to six years old[23]. Therefore, considering this developmental milestone, 30.43% of the patients had delays with speech exchanges not expected for their age.

Estimates indicate that worldwide, 5% to 10% of children under three years of age have some type of communication disorder[28]. However, access to these services is not straightforward despite the growth and increased importance of speech-language services in the SUS over the years[27]. It is known that the wait for speech-language services in the SUS is prolonged, with little user turnover due to longer treatments[6,29]. The continuous demand for speech-language services, as evidenced by the waiting list, underscores the need for resources and strategies to meet this growing need. Many genetic diseases lack available treatment or medication, making support from a multidisciplinary team essential to provide emotional support and guidance, as many patients need to undergo various therapies early on due to the need for motor, sensory, and cognitive stimulation[11]. This is evidenced in the study, as a significant portion of the patients attended a rehabilitation center or received treatment from other health professionals. However, although the SUS guidelines for comprehensive care for patients with rare diseases provide for multidisciplinary care, studies indicate that access to various professionals is still precarious due to both material and human resource shortages and the barriers imposed by the regulation system[11].

Based on the findings of this study, speech-language pathology can play a fundamental role in the early identification, evaluation, and therapeutic intervention to improve communication and quality of life for these patients. As future perspectives, it would be relevant to investigate the effectiveness of speech-language pathology interventions in this group, evaluating how these can aid in the rehabilitation of the communicative abilities of these patients and in their quality of life.

Conclusions

Although only 24% of the patients were diagnosed with a syndrome or genetic condition, it is essential that the genetics team be attentive to warning signs and risk factors for communication disorders. The data presented shows that part of the patients had complaints and/or manifestations in the areas of human communication, especially in relation to language, speech, and deleterious oral habits. Promoting correct and early referral for speech-language pathology is crucial for the application of stimuli that promote the development of various skills essential for child development. Furthermore, it is important to emphasize the need for future research to address the limitations and expand the understanding of the complex interactions between genetics and speech-language pathology.

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