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Stomatognathic and speech alterations are common among children with incontinentia pigmenti

Alterações estomatognáticas e de fala são comuns entre crianças com incontinência pigmentar

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ABSTRACT

Purpose: To identify possible speech-language disorders in children with Incontinentia Pigmenti (IP), seeking to characterize the role of speech therapy in the evaluation and management of this genetic condition. **Methods:** The sample was composed of seven female children diagnosed with IP. **Results:** The patients in the sample had a mean age of 6.4 years. Among the main structural features verified in the patients, highlighted the presence of no physiological diastema and hard palate abnormalities, found both in 85.7 % of the sample, in addition to tooth agenesis in 71.4% of cases. As for functional findings, 71.4 % of the sample had abnormal tongue mobility and 57.1%, inappropriate chewing. As for changes in speech, the main findings consisted of phonetic/phonological alterations, verified in 85.7 % of the sample, being the most common phonetic alteration characterized by distortion of alveolar fricative [s], present in 57.1 % of cases. None of the children had abnormal voice and swallowing according to the used protocol. Furthermore, no detectable hearing abnormality was observed according to claim of the family or by observation during the evaluation. **Conclusion:** In this sample the most frequent speech-language alterations verified among the patients with IP were mainly related to the stomatognathic system structures and speech.

Keywords: Speech, language and hearing sciences; Incontinentia pigmenti; Diastema; Tooth; Palate, hard

RESUMO

Objetivo: Identificar possíveis alterações fonoaudiológicas de crianças com Incontinência Pigmentar (IP), buscando caracterizar o papel da Fonoaudiologia na avaliação e manejo dessa condição genética. **Métodos:** A amostra foi composta por sete crianças do gênero feminino com diagnóstico de IP. Todas foram submetidas aos procedimentos de avaliação nas áreas de motricidade orofacial, deglutição, fala e voz. **Resultados:** Os pacientes que compuseram a amostra tinham média de idade de 6,4 anos. Dentre as principais características clínicas estruturais verificadas, destacaram-se a presença de diastemas não fisiológicos e anormalidades de palato duro, encontradas em 85,7% da amostra, além da agenesia dentária em 71,4% dos casos. Quanto aos achados funcionais, 71,4 % apresentaram alteração de mobilidade da língua e 57,1 %, mastigação inadequada. Em relação às alterações de fala, os principais achados foram alterações fonéticas e/ou fonológicas, verificadas em 85,7% da amostra, sendo mais comum a alteração fonética caracterizada pela distorção na fricativa alveolar [s], presente em 57,1% dos casos. Nenhuma das crianças apresentou alteração de voz e deglutição, de acordo com o protocolo utilizado. Além disso, não se evidenciou anormalidade de audição, de acordo com a queixa familiar ou por meio da observação durante a avaliação. **Conclusão:** Nesta amostra, as alterações fonoaudiológicas mais frequentes entre os pacientes com IP relacionaram-se, principalmente, com as estruturas do sistema estomatognático e com a fala.

Descritores: Fonoaudiologia; Incontinência pigmentar; Diastema; Dente; Palato duro

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INTRODUCTION

Incontinentia pigmenti (IP), or Bloch-Sulzeber syndrome (OMIM #308300) is a rare X-linked autosomal dominant genetic disease[1,2]. It affects mainly female neonates, with significant variations in the clinical manifestation, even in the same family. In most cases, it is considered lethal for male fetuses still in utero[1,3]. However, there are reported cases of male patients with mosaicism for the disease and with Klinefelter syndrome[4].

The estimated frequency of IP is 1 in 50,000 infants. However, this frequency is probably greater, since it can be easily missed, due to the highly variable presentation and lesions that may possibly be confused with other diseases[5]. The cause of IP was attributed to a mutation that inactivates the IKK-gamma gene (IKBKG; OMIM 300248), also known as NEMO, localized in the region 28 of the X chromosome long arm. A positive family history for the disease is found in about 50% of cases[1,6].

The skin manifestations are present in almost all patients with IP and appear in 90% of cases during the second week of life. They are characterized by having four phases: vesicular, warty, pigmentary and atrophic phases, which may follow an irregular sequence and have variable duration[6]. These are usually the first signs observed in IP.

Being a multi-systemic disease[7], other findings, in addition to skin, are frequent. Among these, there are the dental abnormalities, observed in 80% of cases[6] and are mostly tooth agenesis, delayed dentition and conical teeth[2]. Neurological disorders are not uncommon and represent the greatest threat to the quality of life of patients with IP[7]. These include seizures, spastic paralysis, developmental delay and microcephaly[6]. Eye abnormalities are also common, being observed in 35% of cases[2]. Other reported and less common alterations include the involvement of nails, bones, muscles and heart[6-8]. Also noteworthy are the presence of oral abnormalities, high palate, soft palate hypoplasia, cleft lip and palate, congenital hearing loss and cognitive impairment[7].

Given the abnormalities presented by patients with IP, which can arise even after childhood, these patients may show abnormalities of orofacial motor skills, speech, language, hearing and swallowing, indicating the importance of monitoring the speech therapy at the time of assessment, diagnosis and treatment. However, it is noteworthy the lack of related studies. The insertion of speech therapy aims at a better quality of life for IP carriers. It is role of the speech therapist, as a member of the multidisciplinary team, to characterize the manifestations involving language, speech, hearing and voice, as well as the functions of the stomatognathic system, as swallowing and chewing[9].

Currently, advances in health care, both in speech therapy as in Genetics, and multidisciplinary work have attracted increasing interest from both areas, as they evolve as complementary sciences, not only to better understanding of human communication and its disorders, but also for further characterization and development of specific programs for people with genetic syndromes[10].

In this context, the aim of this study was to identify possible speech-language disorders of children with IP, attended in the Dermatology and Genetics Services from the *Universidade Federal de Ciências da Saúde de Porto Alegre* (UFCSPA), seeking to characterize the role of speech therapy in the evaluation and management of this genetic condition.

METHODS

This consisted of an observational and cross-sectional study with prospective data collection. During the study period, we identified 14 patients with IP. The study included only children over the age 2 years due to the focus of clinical assessment and tools for the procedure. We excluded those patients who had comorbidities, such as neurological or progressive neuromuscular diseases, that could affect the results.

From the aforementioned patients, seven were excluded due to contact failure (n=3), different age limits for this study (n=3), or comorbidities (n= 1 patient with myasthenia gravis). Thus, the final sample was composed of seven female children with IP, whose parents agreed to participate in the study and signed a consent form. The study was approved by the Ethics Committee of UFCSPA, protocol number 1723/12.

All children were attended by the Dermatology and Genetic Services from UFCSPA and were evaluated by the speech therapy team, from October 2012 to May 2013.

The patients from this study had skin lesions (Figure 1), diagnosed following the criteria that divide the patients into two groups according to the presence or absence of family history[11]. When there is no IP family history, the presence of at least one major criterion (erythema, vesicles, eosinophilia, typical hyperpigmentation, especially on the trunk, following the Blaschko lines, disappearing in adolescence and alopecic atrophic linear lesions) is required for diagnosis. The minor criteria (dental abnormalities, alopecia, woolly hair, nail abnormalities and retinal disease), when present, support the diagnosis of IP. When there is a positive family history, the presence of any criterion (hyperpigmentation, scarring, linear atrophic lesions with no hair, vertex alopecia, retinal disease, dental anomalies, woolly hair and multiple miscarriages of male fetuses) strongly favors the diagnosis of IP.

Initially, contact was made with the families of IP patients, with the invitation to perform clinical assessment. All patients underwent to the assessment procedures in the following areas: motor orofacial, swallowing, speech and voice. Hearing disorders with previous diagnosis were recorded and observed informally during evaluations. Hearing complaints by patients or family members were registered. For subjective evaluation of neuropsychomotor development delay, the criteria of the *Caderno de Atenção Básica, Saúde da Criança: crescimento*

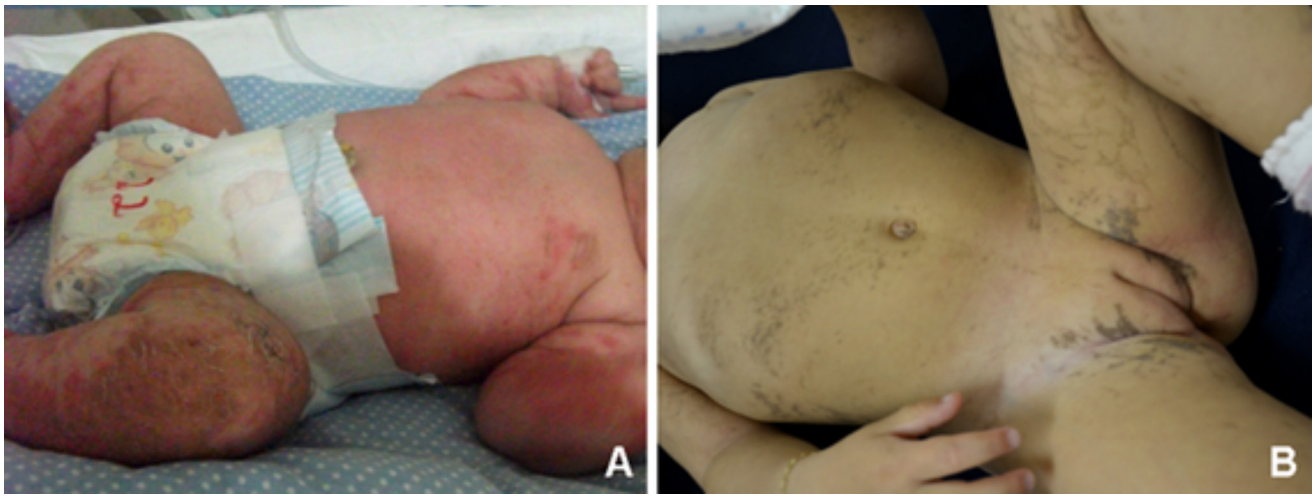

Figure 1. Dermatological findings among patients of the Incontinentia Pigmenti sample. The presence of lesions following Blaschko lines with whorls and bilateral striations, in vesicular (A) and pigmentary phases (B)

e desenvolvimento, Brasília- DF, 2012[12] were considered.

To characterize aspects of independent judges, swallowing, speech and voice of the individuals, a clinical assessment based on protocol MBGR-*Exame Miofuncional Orofacial*[13] was performed. The protocol includes extra and intraoral morphological analyses, assessment of mobility, tone and orofacial sensitivity, as well as breathing, chewing, swallowing and speaking.

For analysis of facial pattern, subjects were classified as Pattern I, II, III, long face or short face. The pattern I is characterized by facial normality and when a malocclusion occurs, this is only a dental malocclusion. Patterns II and III are characterized by positive and negative respectively sagittal step between maxilla and mandible. In long face and short face patterns, the discrepancy is vertical[14]. The analysis, as well as dental characteristics (type of malocclusion, type of teeth, tooth agenesis and non-physiological diastema), were carried out together with the dentists participants of the study[14].

For analysis of food swallowing, solid (French bread), pasty (creamy yogurt) and liquid (water) consistencies were used. It was offered up the food to the patient, and, during that time, cervical auscultation was held and the following aspects were observed: closing lips, tongue posture, food containment, contraction of muscles involved, head movements, noise, coordination and residues after swallowing. In the analysis of speech, it was asked to the patient to count numbers (0-20), days of the week, months of the year, naming pictures involving various sounds and spontaneous speech. Motor coordination was seen in the patient speaks through rate and rhythm. When the child had abnormal speech and this was not clear in the instrument used, it was applied a more specific assessment by the instrument *Avaliação Fonológica da Criança* (AFC) Protocol[15], according to age and the possibility of the individuals in the sample.

Finally, voice analysis was performed requesting the issuance of sustained vowel, where it was observed *pitch, loudness* and voice type, and maximum phonation time through the sustained vowel [a] and sustained consonants [s] and [z]. It is noteworthy that due to the sample heterogeneity in terms of age and socio-cultural characteristics, the evaluations were adapted to the patient profile.

The data analysis was organized in charts and the results expressed as mean and absolute frequency.

RESULTS

The seven patients from the sample were female, aged at assessment moment between 3 years and 7 months and 11 years and 1 month (mean 6.4 years). Only two children (28.6%) had history of preterm birth and in five cases (71.4%), they were born by cesarean section. From the seven patients, two (28.6%) had the mother also affected by IP syndrome.

At the time of the clinical dermatology evaluation, all patients had linear hyperchromic lesions involving mainly the lower limbs; one of them had warty lesions and four had stretch and hypopigmented marks. Other common findings were sparse hair with vertex alopecia (85.7%) and presence of melanocytic nevi (57%).

Among the main extra and intraoral morphological characteristics presented by the patients (Chart 1), we highlight the presence of non-physiological diastema and hard palate abnormalities, found in 85.7% of the sample, and dental agenesis in 71.4% (Figure 2).

As for altered functional findings, we could notice that five children of the sample (71.4%) showed tongue mobility change and four (57.1%) had inadequate chewing (Chart 2).

The main findings related to speech disorders observed in the evaluation were isolated phonetic changes (observed in 57.1% of the sample), being the most common disorder characterized by distortion of alveolar fricative [s]. There was also the presence of isolated phonological alterations and phonetic and phonological concomitant in 14.3% of cases (Chart 3).

Chart 1. Extra and intraoral morphological findings observed in patients with Incontinentia Pigmenti

Structural findings	Patients							Total	
	1	2	3	4	5	6	7	n	%
Facial pattern									
Pattern I	+					+	+	3	42.9
Pattern II		+	+	+				3	42.9
Pattern III					+			1	14.3
Malocclusion									
Posterior cruzade bite	+					+		2	28.6
Anterior open bite		+	+					2	28.6
Increased overbite					+		+	2	28.6
Absent malocclusion				+				1	14.3
Denture									
Deciduous	+	+	+	+	+			5	71.4
Mixed						+	+	2	28.6
Dental agenesis	+			+	+	+	+	5	71.4
Non-physiological diastema									
Presence	+		+	+	+	+	+	6	85.7
Absence		+						1	14.3
Hard palate									
High		+	+	+	+		+	5	71.4
Narrow						+		1	14.3
Normal	+							1	14.3
Asymmetry									
Cheek			+		+			2	28.6
Lips		+						1	14.3
Mandible							+	1	14.3
Chin		+			+			2	28.6

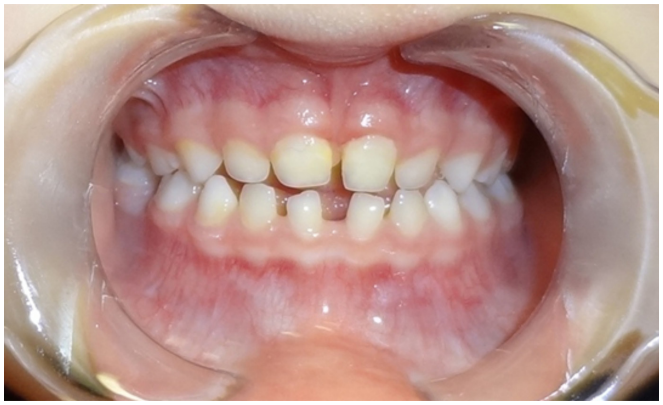

Figure 2. Frontal intraoral image in occlusion showing diastema between lower central incisors and microdontia

None of the children had abnormal voice and swallowing, according to the used protocol. In addition, there was no evidence of hearing abnormality, according to the familiar complaint or through informal observation during the evaluation. All patients in the sample presented neurodevelopment within normal parameters.

DISCUSSION

The prognosis of female patients diagnosed with IP is generally good and depends not only on skin manifestations, but also of extracutaneous features, which may affect the quality of life[16]. The presence of other alterations, such as dental and/or oral abnormalities, are included as minor IP diagnostic criteria[17]. From the patients with the disease, 79.9% have one or more anomalies involving other organs besides the skin. The presence of these anomalies is very important, because, unlike skin changes, they will be present throughout the patient life[18]. Among the minor IP diagnostic criteria[11], we find, in this sample, dental and/or oral alterations. The teeth abnormalities were the most frequent features, including the presence of non-physiologic diastema (85.7%) and tooth agenesis (71.4%), which is in agreement with the findings described in the literature, where dental abnormalities are observed in 80% of the cases, being the most common non-skin IP alterations[6]. The presence of diastema is common in deciduous and mixed dentition in general population. These improve the prognosis for spontaneous alignment of the permanent incisors. According to the literature, 77% of children have dental arch with spacings in deciduous teeth, which remain during the mixed dentition, especially among the upper permanent central incisors[19]. However, the spacing observed in our sample is caused by abnormalities such as micrognathia and abnormalities of dental shape and tooth agenesis, which are part of the IP findings spectrum[19].

As for facial pattern of the patients, the most common were the Pattern I (n=3) and II (n=3). The Pattern I is identified by facial normality, and when a malocclusion occurs, this is only a dental malocclusion. The Pattern II is characterized by a

Chart 2. Functional findings observed in the patients with Incontinentia Pigmenti

Functional findings	Patients							Total	
	1	2	3	4	5	6	7	n	%
Mobility									
Tongue vibration									
Cannot perform	+	+	+	+			+	5	71.4
Attempt to perform					+			1	14.3
Able to perform						+		1	14.3
Oral function									
Breathing									
Nasal	+				+	+	+	4	57.1
Mouth		+	+					2	28.6
Oronasal				+				1	14.3
Chew									
R Unilateral			+			+	+	3	42.9
B Bilateral	+	+			+			3	42.9
L Unilateral				+				1	14.3

Chart 3. Speech disorders observed among the patients in the sample of Incontinentia Pigmenti

Speech disorders	Patients							Total	
	1	2	3	4	5	6	7	n	%
Phonetic									
Distortion in alveolar fricative [s]		+	+	+	+			4	57,1
Distortion in palatal fricative [ʒ]				+	+		+	3	42,9
Distortion in alveolar fricative [z]				+			+	2	28,6
Distortion in palatal fricative [ʃ]			+	+				2	28,6
Distortion in velar fricative [r]					+			1	14,3
Distortion in labial fricative [f]			+					1	14,3
Distortion in alveolar plosive [t]			+					1	14,3
Phonologic									
Fricative anteriorization [ʃ → s]	+							1	14,3
Deletion of non lateral alveolar final liquid [ɾ]	+							1	14,3
Deletion of lateral alveolar initial liquid [l]	+							1	14,3
Consonant cluster reduction			+					1	14,3

positive sagittal discrepancy between maxilla and mandible, i.e., there is a maxillary excess, or a mandibular deficiency[20]. These patients may present tongue interposition during swallowing, tongue on the floor in the rest position and changed chewing (cases 3 and 4)[21]. It was also found bite abnormality and facial asymmetry in 57.1% of cases and high hard palate was verified in 71.4%. In the literature, there are few specific reports of bite changes and facial asymmetry in patients with IP. However, these features could be the result of frequent dental anomalies, which potentially would bring consequences for the form and function of the stomatognathic system[22-24]. Regarding the increased frequency of high hard palate in our sample (71.4%), in a systematic review conducted in the period between 1993-2012, with 1,286 patients diagnosed with IP, where only 513 provided sufficient information to evaluate possible dental and oral anomalies, aged from 1 year, it was observed that 54.4% had some dental and/or oral anomaly, and high hard palate was one of the most frequent oral abnormalities. Despite the relative small absolute number of palate abnormalities, the number was 25 times higher than that found in general population[16], indicating that high hard palate may also be a distinctive and useful finding for the diagnosis, as a minor criterion of IP. One advantage is that it is visible since birth, in contrast to dental anomalies that are detectable only after 1 year[16].

In the evaluation of the stomatognathic system, there was a high prevalence of tongue mobility change (n=6), and oral (n=2) or oronasal breathing (n=1). Chewing was also changed, being predominantly one-sided (n=5). These characteristics can be explained by the close relationship between bone/dental and muscular conditions and functions of the stomatognathic system. The face is an interdependent system and any change in one of its components will result in a general disharmony[23]. In mouth breathers, for example, the tongue tends to remain lower in the oral cavity, which can cause changes in muscle and stomatognathic system functions, including speech production[24].

In the results analysis of the speech evaluation, it was observed that 85.7% of the patients showed abnormalities and

these may be characterized as phonetic or phonological isolated, or even both alterations. Phonetically, 57.1% of patients showed abnormalities, being the most common distortion in fricatives, characterized by an adjustment or compensation used for the production of a phoneme[24]. Changes of phonological origin were less frequent (14.3%) and were also found in children with concomitant phonetic and phonological alterations (14.3%). In the literature, there is no report of speech disorders among patients with IP. Despite the small sample size in our study, the values were higher than those found in general population. In a study of schoolchildren in the city of Belo Horizonte, among the 288 children assessed, 31.9% had some type of speech disorder[25]. In studies developed in Rio Grande do Sul, the prevalence have been ranged from 20.8 to 24.6%[26,27]. The prevalence of phonetic deviation was described as being 18% and phonological disorders were 9.7%. In addition, 4.2% of children had both phonetic and phonological changes[25].

In general, studies report an association between the presence of phonetic deviation and motor orofacial changes[25,28]. Others have shown that the production of /s/, phoneme that showed changes in our sample, requires precise operating conditions and forms of orofacial structures because it has an articulation point more stable and therefore does not allow multiple articulatory trajectories to achieve the same socket. When these conditions are changed, the sound is produced incorrectly and may be distorted[29,30]. Other findings observed in the sample, which may also contribute to the phonetic changes described are the anterior open bite and the increased overbite. The open bite may lead to adjustments in tongue positioning, inserting it, protracting it or raising it[28], as well as the overbite, which reduces the intraoral space due to the decrease of the vertical dimension, which may hinder the necessary movements of the tongue in speech production[24].

Central nervous system abnormalities constitute the most serious complications of IP[2,3]. In our study, none of the patients had abnormal neurological development. Perhaps the small sample size, the criteria adopted for patient selection or their heterogeneity may explain this finding.

CONCLUSION

It could be perceived that speech-language disorders are common among IP patients and they are mainly related to the stomatognathic system structures and speech. It is evident that these abnormalities, especially the phonetic ones, are related to problems of position and mobility of tongue, lips, cheeks and jaw, of dental arch abnormalities and of shape of the teeth. Speech disorders may be considered secondary to the syndrome and may affect the communication development and interaction of these children. In this context, we stress the importance of a multidisciplinary approach, with the inclusion of the speech therapist to the team, for better identification, diagnosis, treatment and monitoring of IP cases, thus enabling an efficient communication and a better quality of life for these patients.

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