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Clinical and speech-language pathology characteristics of neonates hospitalized in a neonatal Intensive Care Unit with suspected genetic disease

Características clínicas e fonoaudiológicas de neonatos hospitalizados em uma Unidade de Tratamento Intensivo neonatal com suspeita de doença genética

Características clínicas y de patología del habla y lenguaje de neonatos hospitalizados en una Unidad de Cuidados Intensivos neonatales con sospecha de enfermedad genética

Chayane Dias Mattos[*]
Melaine Czerminski Larré[*]
Rafael Fabiano Machado Rosa[*]
Paulo Ricardo Gazzola Zen[*]
Sheila Tamanini de Almeida[*]

Abstract

Introduction: Prematurity is a risk factor for the growth and development of neonates. **Objective:** To analyze clinical and speech therapy characteristics of neonates hospitalized in the neonatal intensive care

* Universidade Federal de Ciências da Saude de Porto Alegre, RS, Brazil.

Authors' contributions:
CDM: Assembly of the experiment and data collection, tabulation, writing of the text.
MCL: Data collection and text review.
RFMR: Assembly of the experiment and text review.
PRGZ: Assembly of the experiment, review and writing of the text.
STA: Assembly of the experiment, data collection, tabulation, writing of the text.

E-mail address: Sheila Tamanini de Almeida - sheilat@ufcspa.edu.br
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unit with suspected genetic disease. **Method:** Descriptive cross-sectional study conducted in a hospital in southern Brazil with data collection between November 2020 and September 2021. All neonates who were hospitalized in the ICU attended by the public health system and who were suspected of having genetic etiologies were followed up by the Speech-Language Pathology team. All newborn`s medical records with suspected genetic alterations were analyzed and the medical and the speech-language pathology data were analyzed. **Results:** The sample consisted of 14 premature neonates with a mean gestational age of 36 weeks and 5 days and a mean time of birth, at the time of the speech-language pathology assessment, of 14.6 days of life. Regarding to comorbidities, 71.4% of newborns had some malformation, being multiple in most cases (64.29%). All neonates were using enteral feeding at the time of the speech-language evaluation. At the oral reflexes evaluation it was observed that there was a predominance of patients with a weak rooting reflex and most of them had a present sucking reflex. **Conclusions:** In this study the sample consisted of mainly premature patients who had multiple malformations and all of them used an alternative feeding route, thus suggesting the demand for speech therapy as part of the multidisciplinary care of these neonates.

Keywords: Syndrome; Infant Nutritional Physiological Phenomena; Speech, Language and Hearing Sciences; Genetics, Medical; Infant, Premature; Intensive Care, Neonatal.

Resumo

Introdução: A prematuridade é um fator de risco para o crescimento e o desenvolvimento dos neonatos. **Objetivo:** Analisar as características clinicas e fonoaudiológicas de neonatos hospitalizados na unidade de tratamento intensivo (UTI) neonatal com suspeita de doença genética. **Material e Método:** Estudo transversal descritivo, conduzido em um hospital na região sul do Brasil com coleta de dados entre novembro de 2020 e setembro de 2021. Todos os neonatos que se encontravam internados na UTI, atendidos pelo Sistema Único de Saúde e que apresentavam suspeita de etiologias genéticas foram acompanhados pela equipe de Fonoaudiologia. Foram analisados todos os prontuários dos recém-nascidos com suspeita de alteração genética, extraindo-se os dados médicos e fonoaudiológicos. **Resultados:** A amostra foi constituída por 14 neonatos prematuros com média de idade gestacional de 36 semanas e 5 dias e uma média de tempo de nascimento, no momento da avaliação fonoaudiológica, de 14,6 dias de vida. No que se refere às comorbidades, 71,4% dos recém-nascidos apresentavam alguma malformação, sendo múltiplas na maior parte dos casos (64,29%). Todos os neonatos estavam fazendo uso de via enteral de alimentação durante a avaliação fonoaudiológica. Na avaliação de reflexos orais, observou-se que houve um predomínio de pacientes com reflexo de procura débil, sendo que a maior parte apresentava reflexo de sucção presente. **Conclusões:** Pode-se afirmar que, neste estudo, a amostra foi composta por pacientes principalmente prematuros que apresentavam malformações múltiplas e que todos faziam uso de via alternativa de alimentação sugerindo, assim, a necessidade de atendimento fonoaudiológico como parte da assistência multidisciplinar desses neonatos.

Palavras-chave: Síndrome; Fenômenos Fisiológicos da Nutrição do Lactente; Fonoaudiologia; Genética Médica; Recém-Nascido Prematuro; Terapia Intensiva Neonatal.

Resumen

Introducción: La prematuridad es un factor de riesgo para el crecimiento y desarrollo de los recién nacidos. **Objetivo:** Analizar las características clinicas y de terapia del habla de recién nacidos hospitalizados en la unidad de cuidados intensivos neonatales (UCI) con sospecha de enfermedad genética. **Método**: Estudio transversal descriptivo realizado en un hospital en la región del Sur de Brasil. Todos los recién nacidos que fueron hospitalizados en la UTI y que tenían sospecha de tener etiologías genéticas, fueron atendidos por el equipo de Patología del Habla y Lenguaje. Se analizaron todas las historias clínicas de los recién nacidos con sospecha de alteraciones genéticas, extrayéndose datos médicos y de patología del habla y del lenguaje. **Resultados**: La muestra estuvo constituida por 14 neonatos prematuros con una edad gestacional media de 36 semanas. En cuanto a las comorbilidades, el 71,4% de los recién nacidos presentó alguna malformación, siendo múltiples en la mayoría de los casos (64,29%). Con respecto a los datos de la evaluación de la patología del habla y el lenguaje, todos los

recién nacidos estaban usando alimentación enteral. En la evaluación de los reflejos orales, se observó que hubo un predominio de pacientes con reflejo de búsqueda débil, y la mayoría de ellos tenían presente el reflejo de succión. **Conclusiones:** Se puede decir que en este estudio la muestra estuvo compuesta principalmente por pacientes prematuros, que presentaban plurimalformaciones y que todos utilizaban una vía alternativa de alimentación, sugiriendo así, la necesidad de la fonoaudiología como parte del cuidado multidisciplinario de estos neonatos.

Palabras clave: Síndrome; Fenómenos Fisiológicos Nutricionales del Lactante; Fonoaudiología; Genética Médica; Recien Nacido Prematuro; Cuidado Intensivo Neonatal.

Introduction

Due to the modification and transition from the intrauterine to the extrauterine environment, the neonatal period can be understood as one of the most delicate and difficult moments. The World Health Organization (WHO) definition for preterm infants includes newborns with a gestational age at birth of less than 37 weeks, while those born at less than 28 weeks are considered extremely premature; between 28 and 32 weeks are very premature; and between 32 and 37 weeks are moderate premature.[1,2] It should be noted that the prematurity factor can lead the newborn to intensive care.[3]

It should be noted that prematurity is a risk factor for the growth and development of neonates.[2] In this context, premature neonates in intensive care units (ICU) may have conditions that impact on oral motor skills, such as: physiological and neurological immaturity, respiratory disorders, muscle hypotonia and reduced oral reflexes.[4,5] As a result of these possible alterations, and according to the severity of the case, it is likely that newborns also need an alternative feeding route to obtain the nutrients necessary for their development.[6]

Every year, around 7.9 million children - 6% of global births - are born with serious birth defects, and prematurity and low birth weight have consistently been linked to these conditions.[7] On the other hand, some studies also report a higher frequency of congenital malformations among children born prematurely, compared to those born at 37 or more weeks of gestation.[7,8] In this sense, the Pan American Health Organization (PAHO) defines congenital malformation as any functional or structural anomaly in the development of the fetus resulting from genetic, environmental or unknown factors originating before birth.[9] Even if the birth defect is not apparent and indicates a later clinical manifestation, it is considered a malformation, [7] which, among the many existing ones, include: congenital heart disease, cleft palate and microcephaly among the most frequent. In cases of suspected malformations, a medical genetics team is asked to carry out a genetic evaluation and counseling. Counseling in these cases includes technical clarification and psychological support, through which the patient and family members receive information about the nature and limitations of genetic tests, as well as the benefits, risks and meaning of the results.[10]

In addition to the genetics team, other professionals at the health unit play an important role in welcoming these cases. In this context, speech-language pathologists carry out the evaluation and diagnosis, guidance, therapy - habilitation and rehabilitation - and the improvement of speech therapy aspects of auditory function, language, speech articulation and myofunctional, orofacial, cervical and swallowing systems.[11] Therefore, speech-language pathologists are essential in a multidisciplinary team that works and provides care to newborn patients with malformations, since they may have compromised stomatognathic system (SGS).[12]

Speech-language pathologists are professionals trained to work in a neonatal Intensive Care Unit (ICU) and have knowledge of the anatomy and physiology of the SGS, being able to identify alterations in the sensory-motor-orofacial system, especially with regard to the coordination of sucking functions, swallowing and breathing (SxSxB).[13] Possible compromises of these functions can affect the newborn's feeding relationship, causing a feeding difficulty[5] that will possibly lead the newborn to use an enteral tube to obtain the desired weight gain and necessary nutrients. Thus, the speech-language pathologists will work in the neonatal ICU in order to promote the stimulation of oral feeding in a safe and effective way, instruct and encourage the promotion of breastfeeding, when possible. The speech-language pathologists and the genetics team

can work together in a complementary partnership aiming to allow a diagnosis, prognosis and early intervention in suspected individuals or carriers of genetic alterations.[14]

In this context, this work aimed to analyze the clinical and speech-language characteristics of newborns hospitalized in a neonatal ICU with suspected genetic disease.

Material and methods

This is a descriptive cross-sectional study conducted in a hospital located in the Southern region of Brazil, including data collected between November 2020 and September 2021.

All neonates who were admitted to the ICU, assisted by the Brazilian Unified Health System (SUS) and who had a suspected genetic disease were followed up by the Speech-Language Pathology team. This study was approved by the Research Ethics Committee of the Hospital da Criança Santo Antônio and it was duly registered under the Decision No. 2.489.006. The Informed Consent Form was presented to the guardians of the study participants at the time of evaluation by the Clinical Genetics team.

At first, the researchers analyzed all electronic medical records of newborns with suspected genetic alteration, collecting medical data and speech-language pathology evaluation and follow-up. Next, the researchers created a database detailing a variety of relevant information, such as (i) age in days of life; (ii) gestational age at birth; (iii) birth weight; (iv) Apgar scores; (v) presence and types of malformations; (vi) possible genetic diagnosis; (vii) type of diet; (viii) results of the speech-language pathology assessment; (ix) type of speech-language pathology treatment proposed; (x) defined speech-language pathology strategy; (xi) evidence of improvement in relation to the proposed speech-language pathology treatment; and (xii) need to be referred for speech-language pathology treatment after hospital discharge.

The speech-language pathology assessment performed by the speech-language pathology team of the health unit consisted of a preceptor and a multidisciplinary resident. Data were extracted based on the Premature Infant's Readiness Assessment Instrument for Oral Feeding,[15] which provides objective criteria to indicate the newborn's readiness to initiate oral feeding in a safe and effective manner. This instrument is composed of several parameters, the main ones being corrected age, state of behavioral organization, oral posture, oral reflexes and non-nutritive sucking. It is recommended that a premature neonate reach 28 points on the instrument scale, which varies from 0 to 36, to initiate the transition from tube feeding to oral feeding, with the offer of the mother's breast.[15] The data described in this procedure were used in relation to the genetic evaluation data.

After collection, the data were tabulated and statistical analysis of absolute frequency, mean, median and standard deviation was performed using Microsoft Office Excel® version 2019.

Results

This study included 14 newborns with a mean gestational age of 36 weeks and 5 days and who had 14.6 days of life (SD±17) based on speech-language pathology assessment. Regarding the general characteristics of the neonates, 50% (n=7) of the sample consisted of female neonates; with a mean weight at birth of 2,438g (SD±747.8); approximately 57.14% (n=8) had complications at birth and the median Apgar score Apgar score at the fifth minute was 8. With regard to comorbidities, 71.43% (n=10) of newborns had some malformation, with multiple defects being the most prevalent condition 64.29% (n=9). Regarding heart problems, approximately 71.43% (n=10) of patients had congenital heart disease. As for a possible genetic diagnosis, 42.85% (n=6) had suspected Down Syndrome, while 14.29% (n=2) had suspected Pierre Robin sequence and 14.29% (n=2) suspected 22q11.2 deletion syndrome, also known as velocardiofacial or DiGeorge syndrome. Among the other patients, 28.57% (n=4) had an inconclusive clinical evaluation and/or were awaiting laboratory tests.

As for the speech-language pathology assessment, all newborns were using a tube at the time of the assessment, with 42.85% (n=6) of the newborns using a nasogastric tube, 28.57% (n=4) using an orogastric tube and 28.57% (n=4) were receiving diet through a tube concomitant to the mother's breast. The evaluation of oral reflexes, in turn, showed a predominance of patients with weak search reflex (50%, n=7), and most patients had suction capacity (42.90%, n=6) (Table 1).

Table 1. Results of the Assessment of Oral Reflexes (n=14).

Reflex	Description	N	(%)
Search Reflex	Present	4	28,57%
	Weak	7	50,00%
	Absent	3	21,43%
Sucking Reflex	Present	6	42,90%
	Weak	3	21,43%
	Absent	5	35,71%
Biting reflex	Present	2	14,29%
	Weak	5	35,71%
	Absent	3	21,43%
	Not tested	4	28,57%
Vomiting Reflex	Present	5	35,71%
	Absent	4	28,57%
	Not tested	5	35,71%

Abbreviation: N= Number of newborns.

All neonates underwent oral sensorimotor stimulation after the speech-language pathology assessment and, right after the stimulus, 92.86% (n=13) presented non-nutritive sucking. As for the speech-language pathology strategy, the researchers found that the medical team was instructed to change the orogastric tube for the nasogastric one in 14.29% (n=2) of the cases; while the indication was to maintain the diet exclusively via tube in 57.14% (n=8) of the cases, and the recommendation was to indicate breastfeeding on demand in 28.57% (n=4) of the cases. It should be noted that even with the mother's breast on demand in these cases, the neonates continued with the tube diet.

Finally, with regard to the clinical evolution of the newborn after the treatment performed, an improvement in oral readiness and feeding function was recorded in 57.14% (n=8) of the cases attended, while in 42.85% (n=6) of the neonates there was a demand for speech-language pathology after hospital discharge due to the maintenance of tube feeding.

Discussion

The findings of this study showed that the newborns analyzed can be classified as moderately premature[1,2] and that more than half had complications at birth. According to reports found in the literature, preterm neonates may have many complications and infections that can occur during childbirth or in the postnatal period.[16] Neonatal mortality is higher among preterm infants, and the first months of life are crucial for their neurological maturation.[17] In addition, the vulnerability of premature newborns contributes to the high possibility of risks, injuries and sequelae, such as ventilatory support for a prolonged period, bronchopulmonary dysplasia and jaundice.[18] These complications can directly affect the newborn's development process. Moreover, in addition to this fragility of the premature newborn during the length of stay in a neonatal ICU, there may be consequences, often serious, in relation to the neuropsychomotor and language development of the newborn. It is known that newborns admitted to the NICU are usually constantly manipulated and subjected to various procedures that cause both physical and emotional discomfort. This includes exposure to a stressful environment of noise and light.[2]

Low birth weight and prematurity are known determinant factors related to neonatal mortality[19], and those weighing less than 2,500g are considered low weight.[1] This condition was identified in our sample, adding a greater risk to other comorbidities. Furthermore, there is a study reporting that prematurity and low weight were consistently associated with the presence of malformations[20]. In this sense, there was a predominance of premature neonates who presented malformations and low weight in the sample of this study, according to the cited study.

Congenital heart disease can be defined as an abnormality in the structure or function of the heart[21], being a congenital defect that affects approximately 8 out of 1,000 live births[20]. In this sense, and given that premature neonates are twice as likely to have cardiac abnormalities[20], this study found a predominance of neonates with congenital heart disease, which is in line with reports in the literature. In addition, it is known that neonates with congenital heart disease show a greater predisposi-

tion to bronchoaspiration and the appearance of cyanosis, fatigue and incoordination of suction, swallowing and breathing functions is frequent.[22] It should be noted that neonates with this condition have inadequate biological use of available nutrients, due to the increase in energy expenditure resulting from clinical conditions relevant to cardiac alterations, thus generating a nutritional problem. Furthermore, these neonates may require more time to be fed orally due to cardiopulmonary impairment, which leads to increased effort to feed and fatigue. This feeding difficulty may or may not be associated with oropharyngeal dysphagia.[23]

Regarding the information from the speech-language pathology assessment, all neonates were using a tube at the time of the evaluation. As premature neonates may not be able to feed themselves orally in the first days of life, due to their global and stomatognathic system immaturity[4], this practice can be considered common in this population, requiring the use of alternative feeding methods until they are able to initiate oral feeding.[24] There are reports in the literature that the prolonged use of a probe, in addition to causing a longer hospital stay, can lead to changes in orofacial motor functions, such as delay in coordination between SxSxB, oral hypersensitivity due to little or no oral experience and deprivation of sensory stimuli. These elements can cause difficulty in transitioning from diet to oral feeding and/or rejection of oral feeding, interfering with their oral motor development and delaying hospital discharge.[25]

The beginning of the transition from food to oral route requires reaching some essential milestones, such as current weight, oxygen saturation, degree of clinical stability, maturation process and sucking performance, muscle tone, behavioral organization, control of the environment and posture and coordination to ingest the prescribed diet volume.[14,26] Premature neonates usually show a weak sucking reflex and changes in pharyngeal function, which lead to feeding difficulties.[24] The incoordination of sucking, swallowing and breathing functions[4,24], due to their oral motor sensory immaturity, is one of the most frequent factors of these eating difficulties. Given that sucking is a necessary function for full oral feeding and for proper oral motor development[27] , the premature neonate who presents this difficulty and inability to coordinate may have a negative impact on oral feeding skills.[4] Therefore, it is imperative that the SxSxB functions are coordinated and harmonic. There are some intervention techniques in premature neonates to overcome feeding difficulties, in most cases, caused by prolonged use of a tube. These techniques include non-nutritive sucking and sensory-motor-oral stimulation.[28]

All newborns participating in the study underwent oral sensorimotor stimulation with subsequent development of non-nutritive sucking. This stimulation, which is usually performed in neonates hospitalized in a NICU, aims to develop global maturation, as well as performance and functioning of the digestive system and nutritional aspect.[24] In addition, early stimulation of the stomatognathic system aims to adapt oral functions so that newborns are able to feed themselves orally as soon as possible. Thus, it corroborates with the evolution of the sucking pattern, weight gain and leads to an early hospital discharge.[24] Thus, the performance and speech-language pathology intervention will aim at adapting the oral sensorimotor system and feeding function,[29] in order to improve and develop readiness for oral feeding and efficiency of oral feeding in newborns.

Part of the newborns participating in the study were discharged from the hospital while they were still using a tube, because, despite the improvement and stability in their clinical condition, they still did not have satisfactory development and food security and, as such, did not present conditions for oral feeding. It is important to emphasize that the oral diet should be introduced only when the speech-language pathology assessment confirms that it is safe to do so. Patients who were discharged with an alternative diet were referred for subsequent speech-language pathology therapy

A limitation of this study is the reduced sample size and the lack of comparisons between groups or sample stratification. This study suggests the importance of speech-language pathology practice along with the multidisciplinary team in the premature neonatal population and with genetic diseases. It is also suggested that new studies be carried out with a greater number of premature neonates with genetic diseases and who need speech-language pathology care, as there is a lack of scientific data on this subject and this relationship in the literature. However, the data obtained in this study confirm the findings already described.

Conclusion

The findings of this study allow us to state that there was a predominance of premature patients who had multiple malformations and congenital heart disease. In addition, all participating newborns used an alternative feeding route in the first days of life, most of which were nasogastric. There was also a predominance of patients with a weak search reflex and the strategy indicated for most newborns after the speech-language pathology assessment was to maintain the diet exclusively via tube, thus suggesting the need for speech-language pathology care as part of the multidisciplinary care of these neonates suspected of having a genetic condition.

Due to the alterations described, these patients seem to have a greater predisposition to delayed growth and development if not treated early, requiring follow-up by a multidisciplinary team throughout their childhood and possibly into adulthood.

Finally, there was a need to perform speech-language pathology therapy regardless of having a definitive genetic diagnosis. It should be noted that the diagnosis and the set of clinical findings are essential to carry out a multidisciplinary work and to promote better care for neonates, and thus minimize sequelae. This combination contributes to a greater exchange of knowledge and experiences between both areas, which would improve the care offered to these patients.

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