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# Speech-language pathology aspects in a pediatric case of head and neck arthrogryposis

## *Aspectos fonoaudiológicos em um caso de artrogripose de cabeça e pescoço pediátrico*

### Keywords

Arthrogryposis  
Syndrome  
Deglutition disorder  
Speech-language and Hearing Sciences  
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### Descritores

Artrogripose  
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### ABSTRACT

Arthrogryposis is a rare, multiple, congenital syndrome of non-progressive nature characterized by a series of genetic malformations, as well as stiffness and joint contractures. This is a clinical case study whose objective is to describe speech-language pathology disorders through the evaluation process in a case of arthrogryposis in Pediatrics. The medical records of a patient were analyzed from birth. A complete clinical evaluation of pediatric dysphagia was performed, establishing a diagnosis of severe oropharyngeal dysphagia evidenced by functional and structural impairments. Hearing loss was detected in association with this condition.

### RESUMO

A artrogripose é uma síndrome múltipla congênita rara que se caracteriza por uma série de malformações congênitas e enrijecimento e contrações articulares e não possui caráter progressivo. Trata-se de um estudo de caso clínico, cujo objetivo é descrever a manifestação funcional relacionada à fonoaudiologia através do processo de avaliação em um caso de artrogripose em pediatria. Foi realizada uma análise do prontuário clínico de um paciente desde o nascimento, bem como uma avaliação clínica completa de investigação de disfagia pediátrica, na qual foi estabelecido o diagnóstico de disfagia orofaríngea de grau grave, apontada por alterações no exame funcional e estrutural. Associa-se ao quadro, uma perda auditiva.

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## INTRODUCTION

Arthrogryposis is a rare, multiple, congenital syndrome<sup>(1-3)</sup> of non-progressive nature, characterized by clinical malformations, as well as stiffness and joint contractures. It begins during the period of embryonic development in intrauterine life<sup>(1,2)</sup>.

It is a rare congenital disease - prevalence of 1:300 per 5000 live births, and its clinical symptomatology includes over 150 different disorders<sup>(3)</sup>. There are several types of arthrogryposis, all of which associated with fetal akinesia, that is, decreased fetal movement<sup>(4)</sup>. The most common type of arthrogryposis is the sporadic condition, amyoplasia, which has prevalence of one in 10,000 cases<sup>(5,6)</sup>.

Arthrogryposis is often diagnosed after delivery due to non-observance of fetal movement during the prenatal period<sup>(7)</sup>.

The most common clinical characteristics of this disease are inward bent shoulders (in abduction), knee contractures, elbow extension deformities, flexed fingers and wrists, and dislocation of hips and knees<sup>(8)</sup>. Other possible clinical occurrences are feeding interurrences, which occur because of joint rigidity, a characteristic of arthrogryposis<sup>(1,2)</sup>, affecting the mandible and causing lingual stiffness<sup>(8)</sup>.

Medical and therapeutic monitoring of patients with arthrogryposis should be multidisciplinary since its diagnosis. It includes physicians, geneticists, surgeons, psychologists, social workers, speech-language therapists, and physiotherapists, with all this management and care aiming to provide better quality of life for the patients<sup>(4)</sup>.

Studies addressing arthrogryposis seek to investigate the general characteristics of this malformation. The specific scientific literature does not explore food interurrences, nor does it discuss the features associated with the swallowing function and/or other interferences of the disease with orofacial motricity. This case report aims to describe the functional manifestation related to speech-language therapy through the evaluation process in a case of arthrogryposis in Pediatrics.

## CASE REPORT

This case report was conducted after approval of the Research Ethics Committee of the Hospital da Criança Santo Antônio of the Complexo Hospitalar Irmandade Santa Casa de Misericórdia de Porto Alegre (ISCMPA) under protocol number no. 1900382. The child's guardian signed an Informed Consent Form (ICF) for analysis and dissemination of the data in events and/or scientific journals.

The patient was referred to the specialty outpatient clinic of the Brazilian Unified Health System (SUS) of the Hospital da Criança Santo Antônio, which is part of the ISCMPA, at 17 months of age. Speech-language pathology (SLP) assessment was conducted using the Clinical Evaluation Protocol of Pediatric Dysphagia (PAD-PED)<sup>(9)</sup> due to complaint of swallowing disorder and continuous use of nasogastric tube since the first days of life.

The PAD-PED is a proposal for a clinical evaluation protocol of pediatric dysphagia, based on scientific evidence, which seeks to identify impairments in the swallowing dynamics

of children, considering the stages of development of the stomatognathic system, as well as characterize the clinical signs suggestive of laryngeal penetration and/or laryngotracheal aspiration. This protocol assesses the impact of dysphagia on the functionality of feeding and assists with the most appropriate conduct in the established clinical condition based on the results of the evaluation<sup>(9)</sup>.

The patient underwent neuropsychological development assessment by the Denver II Test, which is a child development risk screening test.

Newborns admitted to the neonatal intensive care unit (Neo-ICU) of Mario Totta maternity hospital, which is part of the ISCMPA complex, are referred to the Neonatal Hearing Screening and the SLP outpatient clinic for audiological assessment. The investigated patient failed the neonatal hearing screening, presenting risk of hearing loss. After the SLP evaluation, the patient was submitted to auditory assessment through conditioned orientation response audiometry, conducted in free-field for pure warble tones using a PA-5 Interacoustics® field audiometer.

This clinical study reports the case of a male infant with previous history of cesarean birth at gestational age of 39 weeks, from the countryside of the state, referred to the Neo-ICU because of presence of congenital fractures and difficulties in breastfeeding. The newborn remained in the Neo-ICU until the age of two months for diagnostic elucidation and clinical condition stability.

In the initial anamnesis, the mother reported very little breastfeeding during the newborn's first days of life, and that when he was admitted at the Neo-ICU, oral feeding was interrupted and nasogastric feeding was introduced. During this hospitalization time, the patient underwent speech-language therapy daily, and his mother was advised not to feed him orally because of risk of laryngotracheal aspiration.

At the first clinical evaluation, conducted on his fifth day of life, during his period of hospitalization at the Neo-ICU, the following information was included in his medical records: flat palate; parted lip posture with raised tongue; absent oral reflexes of searching, biting, and vomiting; weak suction reflex; altered mandible and tongue movement; weak and arrhythmic suction movements with <5 suction/pause pattern; suck-swallow-breathe (S-S-B) incoordination. The oral feeding test with a 1 ml syringe coupled with the gloved finger suction test showed accumulation of milk in oral cavity, with slight anterior leakage.

Still during this hospitalization period, a Newborn and Infant Hearing Screening was conducted through Transient Evoked Otoacoustics Emissions (TEOAE) and Auditory Brainstem Response (ABR), and failing results were observed. At two months of age, the patient was discharged from the Neo-ICU with a referral to genetic evaluation, hearing assessment through Brainstem Auditory Evoked Potential (BAEP-diagnostic), and assistance in SLP outpatient clinic. The BAEP-diagnostic to assess risk of hearing loss was not concluded until 19 months of age due to difficulty in keeping the infant asleep.

The diagnosis of arthrogryposis occurred at 12 months and 27 days of age, with congenital humeral fracture, congenital feet, and craniofacial asymmetry as the clinical features.

**Chart 1.** Sound-source location auditory responses

Minimal response levels (dBNA)	500 Hz	1000 Hz	2000 Hz	4000 Hz
<b>On the right side</b>	30	20	20	20
<b>On the left side</b>	40*	30*	40*	30

\*Sound-source location on the right side; dBNA - hearing level in decibels; Hz - Hertz

At 17 months of age, the patient was admitted to the Speech-language Pathology and Audiology Outpatient Clinic. Auditory reassessment was performed using the PAD-PED protocol, and a diagnosis of severe oropharyngeal dysphagia was established. The following functional and structural impairments were observed: everted lips with parted posture, adequate tonus, and lack of mobility; symmetrical tongue posture resting on buccal floor and reduced mobility; preserved tonus of tongue and cheeks; presence of teeth (maxillary and mandibular central incisors); vocal quality within normality standards verified through crying. Sialostasis, sialorrhoea, and absence of movement or deglutition were observed in the assessment of swallowing of saliva and liquids. Anterior leakage through the labial commissures was observed in the swallowing assessment with liquids. The characteristics of S-S-B incoordination observed in the initial examination remained present in this clinical evaluation. The patient remained tearful throughout the assistance period, which hindered complementation of the testing.

Some delay was observed in the neuropsychological development assessment, with indication of not being able to crawl or sit without support.

At 20 months of age, the patient underwent conditioned orientation response audiometry, whose minimal free-field responses to pure warble tones using a PA-5 Interacoustics field audiometer indicated probable bilateral hearing loss, minimal/mild level in the right ear and higher level in the left ear. Absence of cochleopalpebral reflex to a sound stimulus >90 dB and response to verbal commands (live voice) compatible with level II of Azevedo<sup>(10)</sup> were also observed. The minimal response levels are presented in Chart 1. The audiometric findings also show a delay in auditory function development.

Since 19 months of age, the infant has been under follow-up by the teams of oral and maxillofacial surgery, pediatrics, thoracic surgery, SLP, and physiotherapy; he has been discharged from genetic care.

## DISCUSSION

Studies addressing arthrogryposis seek to investigate the general characteristics of this disease and are associated with stiffness and joint contractures. Our observation is attentive to the speech-language pathology (SLP) disorders resulting from this syndrome, which are not reported in the literature.

The joint stiffness, reported in the literature<sup>(1,2)</sup>, presented by the patient in this case report, with craniofacial and mandibular asymmetry, impairs his orofacial mobility, generating feeding problems, and this difficulty is evidenced in pediatric dysphagia. Dysphagia is defined as any change in the transit of food from

the oral cavity to the stomach<sup>(9)</sup>, and laryngeal penetration and/or laryngotracheal aspiration of food may occur during the feeding process<sup>(11)</sup>.

The established diagnosis of severe oropharyngeal dysphagia characterizes a clinical condition with impossibility of oral feeding because of high risk of presumed laryngotracheal aspiration.

The established clinical condition, resulting from the impairments in the deglutition process presented by the patient because of the characteristics of this disorder, may present disturbance in one of the phases of swallowing, namely, oral, pharyngeal, and esophageal<sup>(12,13)</sup>. In the analyzed case, oropharyngeal impairment of swallowing is evidenced, with changes in posture and orofacial mobility and absence of the protective reflex of coughing. The occurrence of sialoestase and sialorrhoea demonstrates difficulties in the control of saliva, and may require measures of control saliva aspiration.

Indication and use of an alternative feeding route was a safe option for the case at that moment, because enteral nutrition with the use of tubes and/or ostomy is indicated in the impossibility of oral intake<sup>(14)</sup>. Based on the clinical condition of the patient, he was referred to the Pediatric Gastroenterology Service with indication of endoscopic percutaneous gastrostomy owing to the need for enteral diet for a period longer than one month<sup>(15)</sup>.

Results of the auditory assessment and the indication of possible bilateral hearing loss suggest that such condition may be directly associated with the clinical characteristic of muscle stiffening and joint contractures, and this should be elucidated with the supplementation of diagnostic tests through physiological assessment of hearing. In view of the audiological findings, the patient was referred to the Pediatric Otorhinolaryngology Service so that, at medical discretion, such investigations could be resumed.

The patient's discomfort during medical care, persistent crying to the touch, and failed interpersonal interaction can be justified by the constant medical/clinical manipulation, coupled with his delayed neuropsychological development. It is known that hospitalization can determine emotional as well as social harm, and can trigger or intensify the suffering imposed by the change of habits.

## FINAL COMMENTS

The clinical characteristics of arthrogryposis, because of the muscle stiffening and joint contractures that affect the head and neck region, favor the onset of speech-language pathology disorders, triggering structural and functional orofacial impairments, favoring the occurrence of oropharyngeal dysphagia. Hearing loss is associated with this condition.

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#### Author contributions

*JAT*: clinical case analysis, literature review, writing of the manuscript; *JSF*: analysis and preparation of the clinical case, literature review; *LM*: analysis and preparation of the clinical case, literature review; *CL*: clinical case analysis, literature review, revision of references; *LRB*: review of the manuscript, study co-adviser; *MCAFC*: review of the manuscript, study adviser.