Lowe's Syndrome: A case study

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ABSTRACT

This study highlights the complexity of Lowe's syndrome, also known as oculocerebrorenal syndrome (OCR), first described by Lowe in 1952, is a rare genetic condition that primarily affects the eyes, brain, and kidneys. Genetic diagnosis, although possible, is often limited by the costs and slowness of the public health system. Thus, diagnosis is often based on clinical criteria and laboratory tests. The prevalence is estimated at 1 in 500,000 in the general population. The case report presents LG, a child with Lowe's Syndrome with neuropsychomotor delay, congenital cataract and glaucoma. The transdisciplinary team, composed of diverse healthcare professionals, has taken a holistic approach to addressing the complex clinical and therapeutic challenges of the syndrome. The therapeutic interventions focused on improving the quality of life, highlighting the importance of speech therapy and the need for early intervention integrated with a transdisciplinary approach, in addition to the active inclusion of the family in the therapeutic process. The study emphasizes the crucial role of third sector institutions in supporting populations in vulnerable situations and highlights the need for articulation between institutions and the public health system.

Keywords: Oculocerebrorenal Syndrome, Neurodevelopmental Disorders, Early Diagnosis, Interdisciplinary Research, Health Sciences.

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INTRODUCTION

Lowe's syndrome, also known as oculocerebrorenal syndrome (OCR), oculocerebrorenal Lowe's syndrome (OCRL), Lowe's disease, or oculocerebrorenal dystrophy, is a rare genetic condition, first described by Lowe in 1952. According to data from the Lowe's Syndrome Association (LSA), in the United States, there is an estimate of approximately 1 in 500,000 in the general population. This association, which tracks cases in several countries, states that the prevalence is between 1 and 10 affected boys per 1,000,000 inhabitants. Studies in Italy described 34 patients, 33 boys and only one girl, until 2005(1) In Brazil, the descriptions of rare diseases follow the American numbers, because Brazilian statistical data are not sufficient. It is associated with the X chromosome and is transmitted in a recessive manner, resulting from changes in the OCRL-1 gene. This gene plays a crucial role in encoding the enzyme phosphatidylinositol bisphosphate 5-phosphatase. When mutations occur in this gene, an intracellular accumulation occurs that interferes with processes such as intracellular signaling, protein trafficking, and polymerization of the actin cytoskeleton(2).

In 2010, 5 cases of male patients treated at the Federal University of São Paulo(1) were described , and in 2015, there is a report of a male case at the University of Passo Fundo, RS(3) .

This study focuses on the clinical presentation of Lowe's Syndrome, which primarily involves the eyes, brain, and kidneys. Congenital cataracts, nystagmus, and glaucoma are the most frequent ocular manifestations, while renal anomalies often resemble Fanconi syndrome. In addition, neurological manifestations include intellectual deficit, neuropsychomotor delay, and seizures, while the musculoskeletal system is affected by rickets, generalized hypotonia, and weight and height deficiency (4)

While genetic diagnosis is possible, it is often limited by the cost and slowness of public health systems. Thus, the diagnosis of Lowe's Syndrome is often based on clinical criteria and laboratory tests.

In view of this, this case report aims to highlight the importance of early intervention in speech-language pathology, combined with a transdisciplinary approach, in improving the diagnosis and quality of life of the patient and his family. The case of LG, a child with Lowe's Syndrome, presents clinical and therapeutic challenges that illustrate the complexity of this syndrome and the need for comprehensive and coordinated interventions.

PRESENTATION OF THE CLINICAL CASE

For this case study, the ethical principles were complied with (CAE: 79392824.0.0000.5501). The participant's legal guardian signed the Informed Consent Form (ICF), as well as the Image Use

Term, consenting to the use of the photograph without a black stripe, since the ocular characteristic is a determinant for the diagnosis of this syndrome.

The child, LG, arrived at the service, a social organization focused on the care of people with disabilities in the interior of the state of São Paulo, in 2020, at the age of 5 months, referred by the pediatrician due to neuropsychomotor delay, congenital cataract and glaucoma.

At 38 weeks' term birth, cesarean section, the child presented Apgar scores of the first minute = 7 and the fifth minute = 9, without any complications with the mother and the newborn, both were discharged from the maternity ward without any additional guidance.

LG makes up a family of 4 people, father (28 years old), mother (30 years old), older sister (7 years old) and younger sister (5 years old). The father had no family history of illness and the mother had no family history, as she grew up in an adoptive family.

At two months of age, the mother reports that he did not look at her and that his "eyes trembled" (according to the mother), and from this complaint she was referred to the first specialized service, in the capital of São Paulo, for the treatment of congenital cataracts and glaucoma.

The work of this institution, of a transdisciplinary nature, welcomed the child through the social service that inserted him in the agenda of the team composed of: speech therapist, psychologist, physiotherapist, nutritionist, pedagogue, social worker and physical educator.

In the team's evaluation, the general developmental delay, accentuated hypotonia, low weight, short stature and the emotional fragility of the family in the face of so many symptoms were soon clarified.

Thus, the team decided to incorporate the mother, as the main caregiver, in all therapeutic moments with the team, in the opportunity to offer her a welcoming listening, to value her view in the face of daily difficulties, and to provide moments of exchange to find functional means of observation and stimulation in the routine of the service and at home.

Despite her visual difficulties, LG had good relational contact with the staff and children at the institution. Severe hypotonia left him passive for motor activities that always required his mother's help.

In speech-language pathology observation, hypotonia of the orofacial muscles made lip closure difficult, contributed to drooling and inefficient, noisy and oral breathing[5]. Although LG had a good appetite, due to masticatory and postural difficulties, he was still fed with sifted pasty soup and a bottle for water and milk. The food issue was a point of great concern for the family because LG was underweight and the expected height for his age.

In the nutritionist's evaluation, it was found that the menu offered daily was varied and very rich in fiber, proteins and vitamins, therefore, low weight and height could be related to other factors

unknown to the team, but the hypothesis of the inefficiency of the execution of food functions for the absorption of nutrients was raised (6)

The physiotherapy and speech therapy work were carried out in two models: separately and together, weekly, with the presence of the mother. In the unique work of each professional, specific aspects were observed and specific techniques were applied, while in the joint work, the dyad exchanged knowledge for reciprocal complementation, for example, the physiotherapist placed the child in the most comfortable and appropriate posture possible for the speech therapist to offer food, or the speech therapist guided the posture of the oral organs in the body exercises of physical therapy.

The speech therapy work was focused on the organization of oral and eating functions, posture and musculature, within the child's possibilities, so that the moments of liquid and pasty/semi-solid ingestion were as pleasurable and efficient as possible, always associated with language and hearing stimulation. These last two areas were greatly explored with musical activities in groups, with other children from the institution, and individually, in a way that is more directed to communicative language.

The family was guided by the social service regarding their rights and access to health, in carrying out the exams and requesting specialists.

After 10 months of care, we reassessed GL's performance in all areas, and at this moment, at 1 year and 4 months, he sat without support, started crawling, ate solid foods with a varied diet and produced some sounds towards the interlocutor. Regarding his vision, he underwent the surgeries and was adapted to the use of glasses, which contributed greatly to his general development. However, the growth curves remained below average, and the tests requested by the nutritionist still showed important changes. In addition, as he grew up, LG had unique facial features within his family and there was an episode that caught the attention of the team. The child, when leaving the couch, had a fracture of the tibia bone, facts that raised suspicion in the team regarding the diagnosis.

At this time, after months of waiting, the vacancy arose for the consultation with the neuropediatrician, who based on the history, laboratory tests and clinical examination raised the hypothesis of Lowe's Syndrome.

In view of the confirmation of the clinical and genetic diagnosis, the team aligned the therapeutic direction to improve the child's quality of life within the community and family nucleus, since the life expectancy of these people does not exceed the third decade of life.

The speech-language pathology work with the presence of the mother and/or another family responsible (father and paternal grandmother) was made possible by the institution more frequently (two to three times a week), because in addition to the severity of the case, the child presented, every day, more episodes of urinary infection, with many absences.

The direction of the speech-language pathology work maintained the initial objectives of strengthening the orofacial muscles for the functionality of chewing and swallowing functions, which are so important for the development of this child. The removal of the bottle and pacifier suppressed the suction function, but considered as an important instrument in the performance of myofunctional exercises, we adopted, together with the nutritionist, a menu of juices, with a slightly dense texture, to be offered with a straw, in the middle of the morning. The introduction of the straw was very difficult for the child, due to the difficulty in grasping the straw through the lips. With the impossibility of using the straw, even with a large diameter, and the child's refusal to offer the juices in the water intake cup, we moved on to the transition cup with a single hole, which provided slow suction movements, with reduced force, but which facilitated the ingestion of nutritious juices.

The child's communication was efficient from the multimodal point of view[7], he communicated with the mother through small "cries", to say about some discomfort or ask for a lap. To communicate with the therapists, he made the head movements with effort, directing his gaze to the interlocutor and emitting some vowels. From this communicative effort, his speech was always put in evidence to be understood and answered, valuing his communicative act and sharing with his family the possibilities of dialogue that were formed during the interactions.

After 2 years of care, and constant reassessments, photos and filming, we found that the speech-language pathology work should be continuous in cases diagnosed with this syndrome, since during a long period of absence (hospitalizations, COVID, frequent urine infections, among other health complications characteristic of the syndrome) the child returned to present initial complaints and symptoms such as: chewing difficulty, saliva control and excessive body hypotonia.

The work was suspended when the child got a place for kidney surgery in a specialized hospital and due to several health complications, the family opted to move to another city to facilitate the child's care.

DISCUSSION

This case study, focused on Lowe's Syndrome, offers important insights into the care of children affected by this rare and complex condition. Lowe's Syndrome is a genetic pathology that mainly affects the eyes, brain, and kidneys, with clinical manifestations that vary from patient to patient. The case of the patient LG, presented in this study, illustrates the complexity and challenges faced by children with this syndrome and highlights the importance of a transdisciplinary approach in the provision of health care.

Diagnosing Lowe's syndrome can be challenging, as it is a rare condition and many of the symptoms are nonspecific. However, early diagnosis is essential to allow for appropriate

interventions and improvements in the patient's quality of life. In this context, transdisciplinary care plays a key role in early identification and comprehensive treatment.

The child LG was referred for care due to neuropsychomotor delay, congenital cataract and glaucoma. Her case demonstrates how coordination between different health professionals, including speech therapists, psychologists, physiotherapists, nutritionists, educators, social workers and physical educators, can be essential to understand and address the multiple needs of children with Lowe's Syndrome. The transdisciplinary approach allows for a comprehensive assessment of the child's cognitive, motor and social functions, helping to identify areas of intervention needed.

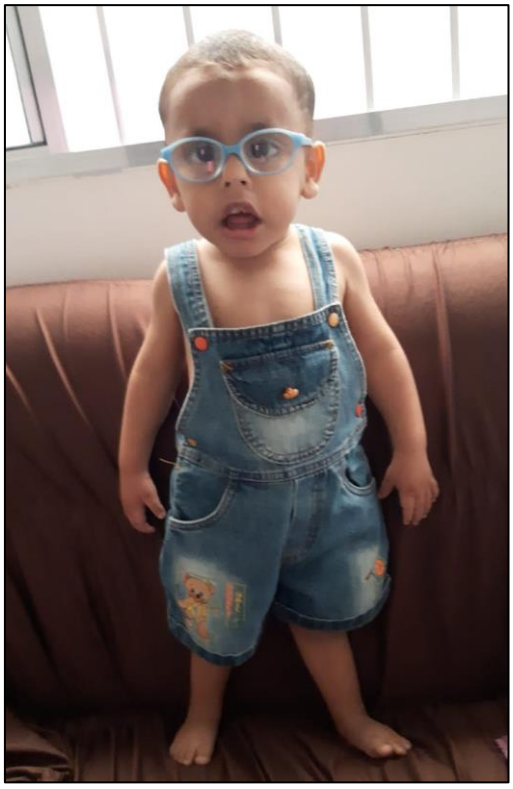
Marked hypotonia, delayed neuropsychomotor development, and vision problems presented by GL are common symptoms in patients with this syndrome. Speech-language pathology monitoring plays a crucial role in improving swallowing function and communication. The strengthening of the orofacial muscles and the introduction of therapeutic strategies, such as the use of straws, help in feeding and promoting the development of language skills.

The case of LG also highlights the challenges faced by the healthcare team due to frequent urinary tract infections and other health complications common in patients with Lowe's Syndrome. The need for continuous interventions is evidenced by the fact that, after periods of withdrawal from treatment, GL presented recurrence of initial symptoms, emphasizing the importance of continuing follow-up.

A relevant aspect is the impact of Lowe's Syndrome on the quality of life of the child and his family. The need for constant interventions, surgeries, and frequent medical care can be emotionally draining. Therefore, the inclusion of the family in the therapeutic process, as active caregivers and participants in therapies, not only offers practical support, but also provides an important emotional basis for the child.

Finally, the emphasis on third sector services [8] and institutions that offer free care to vulnerable populations highlights the need to improve the referral and counter-referral system. Institutions that provide services to vulnerable populations often lack effective support and coordination with other health systems, which can negatively impact therapeutic progress.

This case study highlights the complexity of Lowe's Syndrome and the importance of the transdisciplinary approach [9] in health care delivery. It provides discussion on intervention strategies that can improve the quality of life of children with this syndrome and highlights the need for effective coordination between third sector institutions and the public health system.



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