RELATO DE CASO

PIERRE ROBIN SEQUENCE: A CASE REPORT

A SEQUÊNCIA DE PIERRE ROBIN: UM RELATO DE CASO

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ABSTRACT
The Pierre Robin sequence is characterized by hypoplasia of mandibular area causing micrognathia, glossoptosis and cleft palate in newborns. Its etiopathogenesis is still much discussed in the literature and there is still no consensus on the causal factors of the disease, as well as its treatment, although much discussed, is not fully established. The present article reports a case of a newborn female with the diagnosis of Pierre Robin's Sequence, its evolution until the seventh month of life and planning of therapeutic conduct.
Keywords: Micrognathia. Glossoptosis. Pierre Robin.

RESUMO
O câncer de vesícula biliar é o quinto processo maligno gastrointestinal mais A Sequência de Pierre Robin (SPR) é caracterizada pela hipoplasia de área mandibular acarretando na micrognatia, glossoptose e fissura palatina em recém-nascidos. Sua etiopatogenia ainda é muito discutida na literatura e ainda não há um consenso sobre os fatores causais da doença, assim como o tratamento que não é totalmente estabelecido, embora muito discutido. O presente artigo relata um caso de um recém-nascido do sexo feminino com o diagnóstico de SPR, sua evolução até o 7° mês de vida e planejamento em torno da conduta terapêutica.
Palavras-chave: Micrognatia. Glossoptose. Pierre Robin.

ACESSO LIVRE

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INTRODUCTION

Described in 1923 by Pierre Robin, French dental surgeon, the author's name sequence is characterized by the hypoplasia of the mandibular area that leads to the pathology's triad of classic anomalies: micrognathia, glossoptosis and upper airway alteration, usually caused by cleft palate. Although 90% of cases are associated with a cleft palate, its presence is not mandatory for diagnostic criteria, which requires at least two of the other triad items. The incidence of the disease is 1:8,500 to 1:30,000 births and it is possible that there is a relation to family history.[1,2]

Regarding the presence of anomalies, upper airway obstruction occurs due to involvement of other anatomical structures that may result in difficulty in feeding and respiratory deficiencies, ranging from mild to severe asphyxia. SPR usually comes alone but may also be associated with other syndromes such as: Stickler's Syndrome, myopia caused by retinal detachment and blindness in the first decades of life[4], in 44% of cases; bilateral craniofacial microsomia in 3% of cases; Treacher-Collins Syndrome, an autosomal dominant disorder of craniofacial development that causes mandibulofacial malformation,[5] in 5% of cases and with fetal alcohol syndrome in a lower percentage. There may also be other abnormalities in the auditory system and alterations of nasal and oropharyngeal anatomy.[1,3]

CASE REPORT

Female newborn, daughter of non-consanguineous healthy parents, G1P1A0 mother, without intercurrences in the prenatal period, with the duration of 39 weeks and 1 day and serological tests for HIV, Syphilis, Toxoplasmosis, Cytomegalovirus and Rubella nonreactive. Childbirth was transvaginal, amniorrhexis occurred spontaneously four hours before the expulsive period with clear amniotic fluid without lumps. The Apgar score was 8 in the first minute and 9 in the fifth minute. Weight at birth of 3625g, length of 49cm and head circumference 35cm. Left skin-to-skin for a few minutes, the newborn soon developed respiratory discomfort and used O_2 under a nasal catheter, with partial improvement in saturation. Physical exam showed normal fontanelles, presence of eyeballs, normo-implanted auricular pavilion, microretrognathia, posterior cleft palate, glossoptosis and syndromic facies. Pulmonary and abdominal exams without alterations. Well perfused limbs without edema or malformations.

Patient was referred to neonatal intensive care bed in the use of nasal CPAP. Semi-elemental and hypoallergenic diet, based on extensively hydrolyzed whey protein by nasogastric tube, was prescribed but vomiting persisted. After 2 days, patient had an episode of myoclonic seizure, evolved with severe respiratory failure, was intubated and coupled to mechanical ventilation. Late neonatal sepsis was diagnosed, treatment with broad spectrum antibiotics was given, and the infection improved.

The patient underwent nasopharyngoscopy, which evidenced a retroposition of the dorsum of the tongue with compression of the soft palate against the posterior region of the pharynx and consequent obstruction of the upper airway. Transfontanelle ultrasonography presented hyperechogenicity of the encephalon, probably due to hypoxic-ischemic insult. On the transthoracic echocardiogram, presence of patent foramen ovale, patent ductus arteriosus of small size without hemodynamic repercussion and normal cardiac chambers. 46XX karyotype and abdominal ultrasound with no alterations. Hearing test also without alterations.

During the hospitalization period there were several attempts at extubating without success. After evaluation by pediatric surgery, it was decided to perform tracheostomy and gastrostomy, in addition to correction of gastroesophageal reflux (fundoplication). Patient evolved well in the immediate postoperative period, but 10 days later, presented important abdominal distension associated with worsening of the general condition. The pediatric surgery team decided to do an exploratory laparotomy with gastrorrhaphy due to gastric laceration. There was a satisfactory evolution in the postoperative period and was given diet exclusively by gastrostomy, with good acceptance. Patient was kept hospitalized for another 2 months in a neonatal intensive care unit, with progressive weaning of sedation and oxygen, and was discharged from the hospital with a full diet by gastrostomy and in ambient air.

At the moment, the patient, with age of 7 months, attends pediatric and speech-language pathologist. She did not present any complications since her discharge from hospital. Breathes through tracheostomy and is in the use of infant formula by gastrostomy. oral feeding has been introduced progressively for the last month, according to medical guidance, with good acceptance. Patient is in use of domperidone, ferrous sulfate and vitamin D. At physical examination, patient in good general condition, active, reactive, contacting and with a slight delay in neuropsychomotor development according to the Denver II development scale. Pulmonary auscultation with crackles; cardiac auscultation and abdominal physical examination without alterations. Patient is within the normal curve of length and weight for age, but had low weight gain in the last few months. Once patient reaches a greater weight gain, the surgeon will reevaluate and establish the best definitive conduct for the patient.

DISCUSSION

The first reports of Pierre Robin Syndrome occurred in 1891: two cases of patients with micrognathia, cleft palate and retro glossoptosis reported by Lannelongue and Menard.[6] In 1923, a French dental surgeon, Pierre Robin, described the tendency of the tongue to drop over the hypopharynx, causing airway obstruction, as well as the fissure worsening these children's clinical picture.[7]

In 1966, Latham proposed the concept that such a triad was not a syndrome itself but rather a complex of non-specific

symptoms that occurred in several situations: 1) isolated (not associated with other malformations), 2) as a component of a syndrome, and 3) associated with other developmental disorders that, put together, did not represent any specific syndrome.[8]

Pashayan and Lewis,[9] subsequently suggested the name Pierre Robin Sequence for the triad of craniofacial abnormalities (micrognathia, glossoptosis and cleft palate) due to the fact that they believed it to be a sequential pathogenesis. This denomination has been accepted by many authors and has been widely disseminated in recent years, although the World Health Organization, in its 10th revision of the International Classification of Diseases (ICD-10), still maintains the denomination of Pierre Robin Syndrome.[10]

The incidence of this neonatal pathology varies from 1:2,000 to 1: 30,000 live births, according to available literature.[11] Brazilian data referring to the Hospital for Rehabilitation of Facial Anomalies (Centrinho - Bauru, São Paulo), indicate a total of 1% with PRS among 15,000 patients seen at this facility.[3]

The etiopathogenesis of the Pierre Robin Sequence is still discussed often in literature. The triad that characterizes the sequence has already been experimentally achieved in animals by intrauterine constriction of the mandible resulting in tongue retroposition with obstruction of the cleavage of the palate.[12,13] It is believed that the underdeveloped and retroposed mandible results in a determined sequence of clinical events,[14] in which the retroposition of the mandible predisposes the posterior fall of the base of the tongue in the hypopharynx occluding the airways. However, there is another widely accepted mechanism, which suggests that the tongue is "sucked" into the hypopharynx by the negative pressure generated during inspiration and swallowing.[15] The cleft palate is an aggravating factor that may or may not be present in the congenital malformation of this sequence . It is believed that poor positioning of the tongue, during the embryonic process, prevents medial growth and obstructs the cleavage of the palate, resulting in a U-shaped cleft palate.[15.16]

Some authors believe that heredity is a relevant factor in the etiology of the Pierre Robin Sequence, while others believe that there is no hereditary influence on it.[3,15,17] Several authors evaluated the genetic determination of PRS. In a study by Marques et al,[18] 27.7% of the patients with Pierre Robin Sequence with cleft palate also had a family history with the same alteration, showing a possible genetic origin for these cases. According to these authors, the etiopathogenesis of the sequence would be more related to the presence of the cleft palate than micrognathia, as proposed by some authors.

The manifestations of the clinical picture are due to the triad that characterizes the disease, those being: obstruction of the airways and alimentary difficulties, with the need for nutrition by means of probes frequent in these patients.[3,19,20] This increases the risk of pathological gastroesophageal reflux, as these children are already predisposed to this condition due to negative intrathoracic pressure from respiratory effort.[21] This was observed in the clinical case in question, in which the patient presented with respiratory insufficiency, having to undergo tracheostomy, associated with feeding difficulty, being fed by nasogastric tube, evolving with gastroesophageal reflux and requiring fundoplication surgery for reflux correction and gastrostomy. The manifestations of alimentary difficulties are correlated with the respiratory picture; thus, the treatment of the respiratory obstruction directly implies in improvement of the alimentary picture. There may also be systemic manifestations in 10 to 85% of cases, with skeletal-muscular system alterations being the most frequent.[1.3]

The treatment and the prognosis are based on the severity of the respiratory obstruction, which presents in four types demonstrated through a nasopharyngoscopy, performed in the first months of life. In type 1, the obstruction is due to the retropositioning of the dorsum of the tongue, which comes in contact with the posterior wall of the pharynx. Type 2 promotes obstruction by retropositioning the dorsum of the tongue with compression of the soft palate against the posterior region of the pharynx. This was the type of obstruction presented by the patient in question. In type 3, the lateral walls of the pharynx, when moving laterally, cause obstruction; and finally, in type 4, the contraction of the pharynx is sphincteric, which promotes obstruction. Type 1 is present in 80% of cases and has the best prognosis.[3.22]

The most common types of treatments are: postural treatment (child in prone position), nasopharyngeal intubation (NFI) and surgical procedures (glossopexy, tracheostomy and mandibular distraction).[3] The prone position is effective in simpler cases of obstruction, such as type 1, and it is expected that the child will develop adequate growth of the mandible up to one year of age, which positions the tongue previously leaving the air pathway free. NFI can be used for a period of 15 days, also in cases of type 1 and some type 2, and has effectiveness as definitive treatment in most cases.[23] When there is no improvement with NFI in cases of type 1, it is necessary to perform glossopexy , which involves the fixation of the tongue in the lower lip and mandible.[23,24] The indication of tracheostomy occurs in type 1 cases that did not improve with glossopexy, type 2 cases that were not successful with NFI, and in cases 3 and 4, which are generally extremely severe.[23] Tracheostomy is associated with no significant morbidity and mortality rates.

Mandibular distraction is currently the surgical option of choice in more severe patients and corresponds to the placement of a distractor that promotes mandibular stretching, correcting the posterior position of the tongue and relieving airway obstruction in patients with mandibular hypoplasia. Mandibular distraction can definitively correct micrognathia, eliminate the need for tracheostomy in 90-95% of cases and presents a short postoperative period in intensive care with a low complication rate.[22,25]

In relation to the treatment instituted for our patient, we chose the early tracheostomy due to the patient's inability to breathe from birth, and after several attempts at extubation. The patient still awaits the evaluation of the surgeon to define a definitive treatment that can at least partially correct the abnormalities caused by the syndrome, so that the patient has a better quality of life.

Due to the heterogeneous character of the clinical manifestations in the PRS, there is no consensus in the literature about the best treatment. The individual assessment and choice of treatment should be made by the medical staff accompanying the case. The care of a multidisciplinary team should be involved in the care of a child with PRS in an interdisciplinary way.[3]

CONCLUSION

Although it does not yet have a definite etiology and is in no way a common pathology, PRS does not cause severe damage to the patient's life, provided the diagnosis is made at the time of delivery through investigation of the signs that characterize the triad. The treatment varies according to the case and a gold standard has not yet been defined, however, it is necessary to increase the chances of survival of the child and ends up enabling a life without greater difficulties; because of this, the surgeon's evaluation is necessary whenever the diagnosis is made.

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