Pierre Robin sequence: case report, the relevance of autopsy

Sequência de Pierre Robin: relato de caso, a relevância da autópsia

Cristiano C. Oliveira; Maria Aparecida C. Domingues
Faculdade de Medicina de Botucatu da Universidade Estadual Paulista (Unesp).

ABSTRACT

Pierre Robin sequence is a neonatal disorder characterized by micrognathism, glossoptosis and cleft palate. We reported an autopsy case of a child whose malformations of the oropharynx were identified only at birth. The child was extremely preterm with severe neonatal depression and poor recovery, and the orofacial alterations prevented the correct treatment. There was facial disorder characterized by micrognathia associated with cleft palate and posterior displacement of the tongue, compressing the vallecula, structurally compatible with glossoptosis. This autopsy surpassed the scientific and epidemiological relevance, allowing the family genetic counseling and close monitoring of a subsequent pregnancy.

Key words: pathology; autopsy; Pierre Robin syndrome.

INTRODUCTION

Pierre Robin sequence (PRS) is a neonatal disorder characterized by the triad of micrognathism, glossoptosis and cleft palate. Micrognathia, detected by mandibular underdevelopment, and glossoptosis, identified by the posterior displacement of the tongue, causing obstruction of the pharynx, are consensual aspects of this malformation, constantly described since its first report, in 1923, by the French stomatologist Pierre Robin. In 1934, the same author improved the description pointing out the presence of cleft palate, seen in 90% of cases (1–3). In 1976, this group of malformations was attributed to successive process changes derived from loss of mandibular embryological development (4). This report describes the case of a child, undergone autopsy, whose malformations of the oropharynx were identified only at birth.

CASE REPORT

A 24 year-old woman, 26 weeks pregnant, was admitted to the emergency hospital of Faculdade de Medicina de Botucatu, Universidade Estadual Paulista (HCFMB-Unesp), with clinical manifestations of low back pain radiating to pelvic region, associated with small vaginal blood loss. This was the patient’s second pregnancy. The first pregnancy (about a year before), had ended in spontaneous abortion at 12 weeks. The patient attended prenatal clinical care, and no unusual presentations were reported in her ultrasound examinations. On physical examination, the patient was normotensive, afebrile, ruddy, hydrated and responsive. The obstetric physical examination showed uterine height of 25 cm, cephalic fetal presentation, with good fetal parameters (detectable fetal movements and fetal auscultation of 147 beats per minute). Gynecological physical examination showed full dilation with intact membranes without any signs of lower genital tract infection. The obstetric team diagnosed preterm labor. The patient was hospitalized and underwent a cesarean section.

The child was male, extremely preterm (gestational age of 26 weeks), appropriate for gestational age, with extremely low birth weight. At birth, he had severe neonatal depression with poor recovery (APGAR 1–3–4). The pediatric team observed the presence of orofacial alterations that prevented the accomplishment of tracheal intubation. An emergency tracheotomy was performed. However, after two hours, the child suffered a cardiac arrest and eventually died.
He was transferred to the Pathology Service to undergo autopsy. The fetus weighed 825 g (average reference value: 739 g) and measured 32.2 cm (average reference value: 32.2 cm). The head, chest, and abdominal girth measured 23.5 cm, 21 cm, and 18 cm, respectively. The right and left feet measured 5 cm each. Brain, heart-lung block, liver, spleen, right kidney, and left kidney weighed respectively 110 g (average reference: 105 g), 85 g (average reference value: 25.8 g), 45 g (average reference value: 32.1 g), 5 g (average reference value: 1.5 g), 5 g (average reference value: 7 g) and 5 g (average reference value: 7 g). The placenta was not sent for pathological examination.

At ectoscopy, there were the tracheostomy hole and umbilical catheterization. In addition, there was facial disfigurement characterized by micrognathia (Figure 1A). The inspection of the oral cavity showed cleft palate (Figure 2), more pronounced in the posterior portion. After the removal of organs, the block of the cervical viscera pointed to posterior displacement of the tongue, compressing the vallecula, structurally compatible with glossoptosis (Figure 1B). No other alterations were detected in the organs.

The malformations observed at autopsy were consistent with those observed in PRS. Preterm birth of the child was an important factor in the outcome of the case: the patient had severe neonatal depression, requiring tracheal intubation, a procedure that was not possible due to the set of orofacial problems. Although tracheostomy was attempted, the baby survived for only two hours, progressing to respiratory failure, terminal cause of death.

The autopsy proved to play an essential role in the case, as malformations were not detected before birth. The family could undergo genetic counseling at a specialized service. In the subsequent pregnancy, the third one, two years after the child presented in this report, mother and fetus were closely monitored by a high-risk obstetric care service, ensuring periodic morphological ultrasound to screen for alterations.

**DISCUSSION**

The incidence of PRS is estimated at 1:8,500-14,000 births, and this disease is associated with high morbidity due to the airway obstruction provided by poorly set formations of the oropharynx\(^5\). Epidemiological studies show mortality rates between 1.7% and 11.3% over ten years, emphasizing that, in 26% of the cases, there is the coexistence of some other congenital diseases\(^3\).

The PRS can present in three different ways: as part of another known syndrome; associated with other changes without composing a specific syndrome; or in isolation, unassociated with other significant findings. The isolation, as noted in this report, occurs in 40% of the cases\(^6\).

Among the syndromes that have PRS as a component, the most commonly reported is Stickler syndrome, which demonstrates changes in the eyes, ears, and skeletal system. Manifestations are not always apparent at birth\(^2\). Other reported syndromes are velocardiofacial syndrome (associated with mutations in the 22q11.2), alcohol-fetal syndrome, and trisomy 18\(^3\). Cardiac or limb abnormalities have also been reported in PRS.

PRS generally presents with cleft palate, but the prognosis tends to be positive, even with clinical outcomes\(^3\). Patients with PRS in its isolated form have feeding and breathing difficulties due to airway obstruction. The main manifestations are shrill breath, recurrent
RESUMO

Sequência de Pierre Robin é uma doença neonatal caracterizada por micrognatismo, glossoptose e fenda palatina. Relatamos um caso de autópsia de uma criança cujas malformações da orofaringe somente foram identificadas ao nascimento. A criança era pré-termo extremo com depressão neonatal grave e má recuperação, e as malformações orofaciais impediram o correto tratamento. Havia alteração facial caracterizada por micrognatia associada a fenda palatina e posteriorização da língua, estruturalmente compatível com glossoptose. A autópsia extrapola as relevâncias científica e epidemiológica, permitindo o aconselhamento genético familiar e o monitoramento adequado da gestação subsequente.

Unitermos: patologia; autópsia; síndrome de Pierre Robin.

REFERENCES


