Pierre Robin Syndrome: evaluation of a large series of patients treated in two Brazilian centers

Síndrome de Pierre Robin: avaliação de uma grande série de pacientes tratados em dois centros brasileiros

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RESUMO

Introdução: Sequência de Pierre Robin (SPR) é uma importante causa de distresse respiratório após o parto. Apesar dos avanços no tratamento, ainda existem controvérsias quanto à melhor abordagem nos recém-natos com SPR. Foi realizada uma revisão do tratamento e dos resultados de pacientes com SPR nos últimos 12 anos. **Método:** Foram identificados os pacientes com diagnóstico clínico de SPR em 2 centros brasileiros de fissurados (Centro de Atendimento Integral ao Fissurado Lábio Palatal - CAIF e Unidade Craniofacial – Hospital de Clínicas da Universidade de São Paulo - USP) e coletados os dados. **Resultados:** Foram identificados 51 pacientes. A posição prona e tubo nasofaríngeo foram os primeiros métodos de tratamento instituídos para manutenção da via aérea, com taxa de sucesso de 74,5%. Intervenções cirúrgicas foram necessárias em 25,5% dos pacientes devido à falha do tratamento conservador. As técnicas cirúrgicas utilizadas foram a distração mandibular (4 casos), traqueostomia (4 casos) ou ambas (5 casos). O tratamento para a dificuldade de alimentação consistiu de manobras de posicionamento, modificação dos bicos das mamadeiras, sonda naso ou orogástrica e gastrostomia. **Conclusão:** O tratamento conservador (posição prona e tubo nasofaringeo) deve ser o primeiro método a ser instituído nesses pacientes. A distração osteogênica deve ser a primeira opção cirúrgica e substitui a traqueostomia e glossopexia em casos mais graves.

Descritores: Síndrome de Pierre Robin. Fissura palatina. Osteogênese por Distração.

ABSTRACT

Background: Pierre Robin Sequence continues to be an important reason of respiratory distress after the delivery. Besides advanced knowledge in its treatment, controversies exist about the best approach to newborn with PRS. We reviewed patients with PRS treated last 12 years, focusing the treatment and outcome. **Methods:** Patients with a clinical diagnosis of PRS were identified. Data were collected. **Results:** 51 PRS patients were identified. Prone position and nasopharyngeal airway stent were our primary method of airway management, and had 74.5% of success rate. Operative intervention was undertaken for failure of non-operative treatment methods in 25.5% of patients. Surgical techniques utilized were mandible distraction (4 cases), tracheostomy (4 cases), or both (5). Treatment methods to address feeding difficulty included upright feeding techniques, modification of the nipple for bottle feeding and temporary use of a nasogastric or orogastric feeding tube. **Conclusion:** Conservative treatment (prone position and nasopharyngeal tube) should be the first approach for PRS. Distraction osteogenesis may replace tracheostomy and tongue-lip adhesion in more severe cases.

Key words: Pierre Robin Syndrome. Cleft palate. Osteogenesis, Distraction.

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INTRODUCTION

Pierre Robin sequence (PRS) includes respiratory distress due to micro/retrognathia and glossoptosis[1,2]. Risk to die in the first moments of life exists. The etiology is so complex, and may be due to intrauterine overpressure over the mandible, pharmacological toxicity, or genetic error[3]. PRS may occur isolated or associated to other anomalies[4]. Many therapeutic modalities, conservative and surgical, have been described There is great discussion about the treatment of airway obstruction. Majority of the infants does not need of surgical treatment[3]. High level of success using prone position has been published. Other non-surgical options were also described, as nasopharyngeal tube with non-consistent results[5].

Tongue-lip adhesion, mandible distraction and tracheostomy are utilized worldwide to treat more severe cases[4]. The first surgical approach was tracheostomy, which bypasses the site of obstruction and is highly effective. However, number of complications for the patients is so numerous that it is used in extreme situations[6].

The bilateral mandibular distraction will resize the jaw to the ideal and consequently pushing the tongue forward. It has provided definitive correction of both airway obstruction and micrognathia. The cost to patients and families appropriately treated by mandibular distraction osteogenesis is substantially less than tracheostomy[7,8]. Nowadays, there is a preference to indicate distraction osteogenesis as the standard treatment to airway obstruction; nevertheless this procedure is not free of morbidity[6,9].

Due to doubt about the best treatment for these infants with PRS, we run the retrospective study in two Brazilian cleft centers, aiming to evaluate of a large series of sequential patients.

METHODS

This is a retrospective study in two Brazilian cleft centers (Assistance Center for Cleft Lip and Palate – CAIF, and Craniofacial Unit – Hospital das Clínicas of São Paulo University – USP). Patients with a clinical diagnosis of PRS were identified at each institution, between 1996 and 2008.

Data were collected including: gender, date of birth, diagnostic modalities, treatment and age at which it was rendered, follow-up time, complications, and outcomes.

RESULTS

We identified 51 cases of PRS in the period. There were 60.8% females and 39.2% males; 17.64% of the PRS patients had an identified syndrome, as Stickler, Moebius, Goldenhar, Richieri Costa, Hanhart, Cornelia Lange syndromes and cleft #30 (Table 1). Several anatomical abnormalities were identified, including 96% of patients with micrognathia, in varying degrees, 90.2% with associated cleft lip and palate, and glossoptosis in 66.6%. Respiratory distress was identified in 60.8% (Table 2).

The management of airway obstruction was first treated with conservative methods such as prone position and nasopharyngeal airway stent (Figure 1), having 74.5% of success rate and being our primary method of airway management. Operative intervention was undertaken for failure of non-operative treatment methods in 25.5% of patients. Mandible distraction (Figure 2) was utilized in 30.7% of these cases, tracheostomy in 30.7%, and tracheostomy followed by DO in 38.4% (Table 3). We did not indicate any case to tongue-lip adhesion. Four out of five patients had their tracheostomy performed previously, out of our centers. Feeding difficulties (54.9%) were managed using upright feeding techniques, modification of the nipple for bottle feeding, temporary use of a nasogastric or orogastric feeding tube and placement of a gastrostomy.

Table 1 – Associated Syndromes to PRS.

Associated Syndromes	Cases	%
Stickler	3	5.88%
Moebius	1	1.96%
Goldenhar	1	1.96%
Richieri Costa	1	1.96%
Hanhart	1	1.96%
Cornelia Lange	1	1.96%
Cleft #30	1	1.96%
Total	9	17.64%

Table 2 – Abnormalities in PRS.

Type	Cases	%
Micrognathia	49	96.07%
Cleft lip and palate	46	90.19%
Glossoptosis	34	66.66%
Airway distress	31	60.78%
Feeding difficult	28	54,90%

Figure 1 – Patient with Stykler syndrome, treated with nasopharyngeal tube during 21 days. Post-operative picture with 2 years of age.

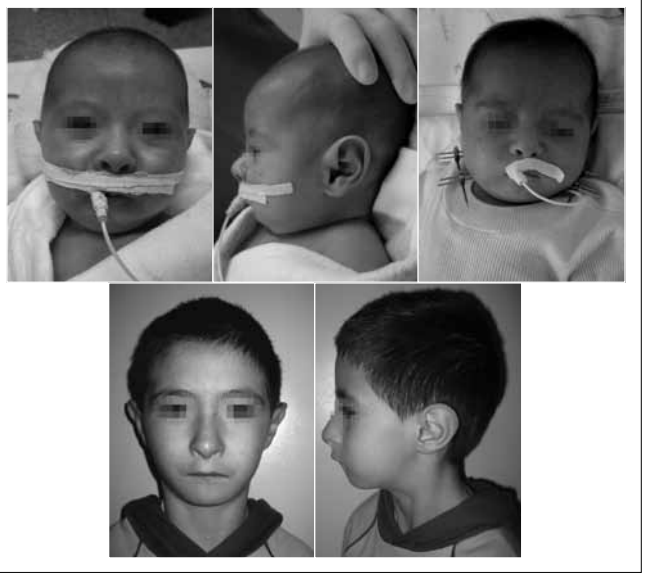

Figure 2 – Patient with Stykler syndrome, treated with mandible distraction after 2 months of NPT. Post-operative picture with 5 years of age.

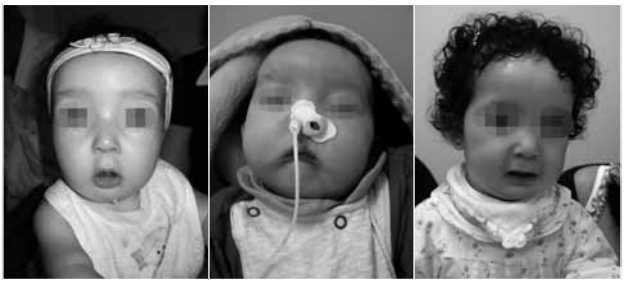

Table 3 – Surgical procedures.

Surgical Procedures	Cases	%	In	Out
Mandible distraction	4	30.76%	4	—
Tracheostomy	4	30.76%	1	4
Both (tracheostomy + distraction)	5	38.46%	5	—
Tongue-lip adhesion	—	—	—	—
Total	13	25.49%	11	2

In: treatment performed in our center; Out: previous treatment in other center

DISCUSSION

PRS was described as a respiratory distress caused by a glossoptosis and micrognathia, but beyond this problem also causes difficulty in eating and talking in varying degrees[1,2]. This syndrome has a frequency of 1:2000 to 1:50.000 births and is an uncertain etiology (intrauterine pressure directed in the jaw, pharmacological, toxic, sporadic and genetic)[1,3]. Cleft palate is often associated with PRS; a small mandible, malposition the tongue in a vertical position, obstructing midline union of the migrating palatal shelves and resulting in a palatal cleft. Some authors do not strictly include cleft palate in their definitions of PRS and this situation should have the timing and methods of repair should independent of airway procedures[3]. Furthermore, this sequence may be associated with several other syndromes such as Stickler, Moebius, Goldenhar, Richieri Costa, Hanhart, Cornelia Lange syndromes and cleft #30, and may delay the diagnosis[4]. The airway obstruction of PRS may cause inability to feed, failure to thrive, dehydration, exhaustion, electrolyte imbalance, cor pulmonale and death. A number of therapeutic maneuvers have been recommended to overcome this obstruction, since conservative methods to invasive situation[1]. The non-surgical options include prone position, nasopharyngeal tube, no oral nutritional support and respiratory support[1].

Some authors reported that treatment with positioning is not as effective for control of airway obstruction in newborns due to difficulty in maintaining the position of these children[5]. There is the question as to monitor these children submitted to conservative treatment and the need for prolonged hospitalization[10]. However several other authors are in favor of the use of prone position for the first treatment modality. The justification is based on the majority of patients with PRS usually presents a mild obstruction and free of morbidity inherent to procedure[3].

The use of nasopharyngeal tube is also a good option for the correction of respiratory distress and enables greater security and comfort for the patient and family, making it possible for them to manage and care for their children in a safer manner[5].

We obtained around 74% of positive results, with improvement of respiratory distress, using conservative methods. Most patients were nonsyndromic, and possible discharge from the hospital earlier.

In our opinion, surgical treatment is reserved for cases where there is failure of conservative treatment because the morbidity of the procedure[7]. In literature, the most commonly used options are glossopexy, mandibular distraction, and tracheostomy. The glossopexy was proposed in 1911 and only accepted in 1940. This technique has some advantages such as efficacy in the management of respiratory distress and less necessity for special hospital care and, for some authors, is the initial treatment in this situation[7,11]. However, posterior traction of the anterior region by the tongue-lip adhesion prevents the anteroposterior growth of the jaw.

There are some doubts about the best treatment for severe airway obstruction associated with difficulties to sleep and maintenance of oxygenation. The "gold standard" for alleviating airway obstruction, including those in neonates and infants, has been the tracheostomy[8]. However, even though the procedure may be lifesaving, it has been associated with complications and developmental impacts. Children with tracheostomies remain in the hospital much longer than other children[9]. Tracheostomy-related mortality rate of 0.5% and a complication rate of 19% in the first week following the procedure[7,8]. They also reported a late complication rate of 58%, all of which had an impact on clinical outcomes (like tracheal stenosis, pneumothorax, tracheal tube obstruction). Moreover, the tracheostomy needs a daily special care and impaired speech and language development[2,8]. There was a direct correlation with age of the patient at the time of the procedure (more deleterious in young children), length of tracheal cannulation, and future speech and language development. Additionally, tracheostomy may also impair normal feeding and swallowing mechanisms. This high complications and mortality levels are the main reasons that we avoid to use tracheostomy[4].

We had a patient who came to our center already with the tracheostomy, which during the preparation for surgery, at home, died due to obstruction of the cannula. Although it has these disadvantages and it is not benign procedure, this method is more effective for the management of air and ends up being reserved for more severe cases[7].

Mandibular distraction is one of the options to treat in severe situations and it will solve the air obstruction in early post-operative. The bilateral mandibular distraction just resizing the jaw to the ideal and consequently pushing the tongue forwardly[8]. Because the process of distraction is gradual and controlled, the distracted bony segment has the capacity to lengthen its primary callus, at the same time allowing the surrounding composite soft tissues to be "recruited" or "stretched" simultaneously[8]. Kaban et al. have successfully demonstrated myocyte proliferation in the masseter muscle, "exuberant" periosteal osteogenesis, and expression of multiple growth factors when histologically evaluating the distracted mandible[3,8]. Some children with PRS will have mandibular ''catch-up'' growth in the first year of life, treatment options with an absolute minimal morbidity should

be developed[8]. Most mandibular distraction associated with complications are minor and correctable (e.g., fracture of the transport segment, difficulties in finishing osteotomy, incorrect direction of distraction, suture dehiscence, mucosal perforation, and bone formation defects)[8]. A few rare, but significant complications have also been reported like pain not related to the operation, functional disturbances in the movement of the jaw, weight loss, temporary unilateral facial nerve palsy, and transient unilateral hypoesthesia of the inferior alveolar nerve e temporomandibular joint disfunction[11,12]. Even it is used in older children for correction of non-symmetries, it is not our first choice for treating air in PRS. The surgical procedures have advantages on the management of the airway, but neither is free from complication[7,8,13-15].

CONCLUSION

The conservative treatment (prone position and nasopharyngeal tube) should be the first approach for PRS, with great number of patients managed satisfactorily. Mandibular distraction may replace tracheostomy and tongue-lip adhesion in more severe cases.

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