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Pierre Robin Syndrome

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Abstract
Pierre Robin sequence (PRS) is classically described as a triad of micrognathia, glossoptosis, and airway obstruction. Infants frequently present at birth with a hypoplastic mandible and difficulty breathing. The smaller mandible displaces the tongue posteriorly, resulting in obstruction of the airway. Typically, a wide U-shaped cleft palate is also associated with this phenomenon. PRS is not a syndrome in itself, but rather a sequence of disorders, with one abnormality resulting in the next. However, it is related to several other craniofacial anomalies and may appear in conjunction with a syndromic diagnosis, such as velocardiofacial and Stickler syndromes.
Infants with PRS should be evaluated by a multidisciplinary team to assess the anatomic findings, delineate the source of airway obstruction, and address airway and feeding issues. Positioning will resolve the airway obstruction in ~70% of cases. In the correct position, most children will also be able to feed normally. If the infant continues to show evidence of desaturation, then placement of a nasopharyngeal tube is indicated. Early feeding via a nasogastric tube may also reduce the amount of energy needed and allow for early weight gain. A proportion of PRS infants do not respond to conservative measures and will require further intervention. Prior to considering any surgical procedure, the clinician should first rule out any sources of obstruction below the base of the tongue that would necessitate a tracheostomy. The two most common procedures for treatment, tongue–lip adhesion and distraction osteogenesis of the mandible.

Keywords: Pierre Robin sequence, micrognathia, glossoptosis, airway obstruction, distraction osteogenesis

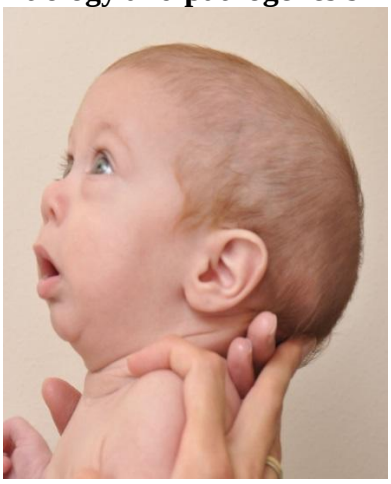
Introduction

Lannelongue and Menard are the first to described Pierre Robin syndrome in 1891 in a report on 2 patients with micrognathia, cleft palate, and retroglossoptosis. In 1926, Pierre Robin published the first case of an infant with the complete syndrome. Until 1974, the triad was known as Pierre Robin syndrome; however, the term syndrome is now reserved for those errors of morphogenesis with the simultaneous presence of multiple anomalies caused by a single etiology. The term sequence has been introduced to include any condition that includes a series of anomalies caused by a cascade of events initiated by a single malformation.

Definition of Pierre Robin syndrome

Pierre Robin syndrome (abbreviated to PRS, and also known as Pierre Robin sequence, Pierre Robin malformation, Pierre Robin anomaly or Pierre Robin anomalad) is a congenital condition of facial abnormalities in humans. PRS is a sequence, i.e. a chain of certain developmental malformations. The three main features are cleft palate, retrognathia (abnormal positioning of the jaw or mandible) and glossoptosis (airway obstruction caused by backwards displacement of the tongue base).

Etiology and pathogenesis



Incidence

This heterogeneous birth defect has a prevalence of approximately 1 per 8500 live births. The male-to-female ratio is 1:1, except in the X-linked form.

Etiology

- ✓ Autosomal recessive inheritance is possible.
- ✓ An X-linked variant has been reported involving cardiac malformations and clubfeet.

Pathogenesis

Three pathophysiological theories exist to explain the occurrence of Pierre Robin sequence.

- The mechanical theory: This theory is the most accepted. The initial event, mandibular hypoplasia, occurs between the 7th and 11th week of gestation. This keeps the tongue high in the oral cavity, causing a cleft in the palate by preventing the closure of the palatal shelves.
- This theory explains the classic inverted U-shaped cleft and the absence of an associated cleft lip.
- Oligohydramnios could play a role in the etiology since the lack of amniotic fluid could cause deformation of the chin and subsequent impaction of the tongue between the palatal shelves.
- The neurological maturation theory: A delay in neurological maturation has been noted on electromyography of the tongue musculature, the pharyngeal pillars, and the palate, as has a delay in hypoglossal nerve conduction. The spontaneous correction of the majority of cases with age supports this theory.
- The rhombencephalic dysneurulation theory: In this theory, the motor and regulatory organization of the rhombencephalus is related to a major problem.

Otolaryngologic manifestations

- ❖ 91.7% reported cases of microganthia (majority)
- ❖ Glossoptosis is noted in 70-85% of reported cases.
- ❖ Macroglossia and ankyloglossia are relatively rare findings, noted in 10-15% of reported cases.
- ❖ Nasal deformities are infrequent and consist mostly of anomalies of the nasal root.
- ❖ Dental and philtral malformations occur in one third of cases.
- ❖ Laryngomalacia occurs in approximately 10-15% of patients with Pierre Robin sequence.
- ❖ Gastroesophageal reflux and esophagitis has also been described.

❖ Speech defects occur frequently in patients with Pierre Robin sequence. Velopharyngeal insufficiency is usually more pronounced in these patients than in those with isolated cleft palate.

Systemic manifestations

In general, systemic anomalies are documented in 10-85% of reported cases.

- Ocular anomalies are reported in 10-30% of patients.
- Cardiovascular findings such as benign murmurs, pulmonary stenosis, patent ductus arteriosus, patent foramen ovale, atrial septal defect, and pulmonary hypertension have all been documented. Their prevalence varies in the literature from 5-58%.
- Anomalies involving the musculoskeletal system are the most frequent systemic anomalies (noted in 70-80% of cases). They frequently include syndactyly, dysplastic phalanges, polydactyly, clinodactyly, hyperextensible joints, and oligodactyly in the upper limbs.
- Central nervous system (CNS) defects such as language delay, epilepsy, neurodevelopmental delay, hypotonia, and hydrocephalus may occur. The incidence of CNS defects is around 50%.
- Genitourinary defects may include undescended testes (25%), hydronephrosis (15%), and hydrocele (10%).
- Associated syndromes and conditions include Stickler syndrome, trisomy 11q syndrome, trisomy 18 syndrome, velocardiofacial (Shprintzen) syndrome, deletion 4q syndrome, rheumatoid arthropathy, hypochondroplasia, Möbius syndrome.

Diagnosis

- The syndrome is generally diagnosed clinically shortly after birth.
- The infant experiences respiratory difficulty, especially when supine.

Conservative management

The goals of treatment in infants with Robin sequence focus upon

- Maintenance of patency of airway
- Optimum nutrition for proper growth and development

a) Airway Compromise

- Prone positional therapy has proved to be highly efficient in airway management. (63% of infants responded to prone positioning**.** (Smith and Senders, 2006, Int J Pediatr Oto).
- Oral airway placement, laryngeal mask, nasopharyngeal stenting, and short-term intubation (< 2 wk) are other options.

b) Feeding difficulties

- Gastroesophageal reflux (GERD) seems to be more prevalent in children with Robin sequence (Dudkiewicz, March 2000, CPCJ) specifically by worsening airway obstruction.
- Remedies include - Upright feeding techniques, modification of the nipple for bottle feeding, temporary use of nasogastric or orogastric feeding tube, and the placement of a gastrostomy, small and frequent feedings (to minimize vomiting), and/or pharmacotherapy (such as proton pump inhibitors).

Surgical management

Infants with pronounced micrognathia may experience severe respiratory distress or failure to thrive. Treatment is prioritized according to the severity of airway compromise followed by the extent of feeding difficulties.

- Tracheostomy remains the most widely used technique. Other surgical procedures, such as subperiosteal release of the floor of the mouth (see the image below), and different types of glossopexy, such as the Routledge

procedure or other forms of tongue-lip adhesions, can be used.

- Mandibular lengthening by gradual distraction may be used for severe mandibular hypoplasia that causes obstructive apnea. [Paes et al (2013) compared the outcome and cost-effectiveness of tracheotomy versus mandibular distraction. They demonstrated that tracheostomy was 3 times more expensive and indirect costs were almost 5 times higher. They also reported 4 times more complications with tracheostomy.]

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