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Pierre Robin Syndrome: A Review

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Abstract-
Pierre Robin syndrome (PRS) is defined as a triad of small jaw, tongue retraction, and air passage hindrance, wherein infants often exhibit an immature mandible and respiration difficulties at birth. The small mandible drives the tongue backward, leading to PRS. In general, a cleft palate which is broad and U shaped is additionally related with this anomaly. PRS is not a syndrome alone, though instead various disorders, with single anomaly leading to another. Nevertheless, it is related to many other craniofacial abnormalities and may emerge together 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. 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.

Keywords: Glossoptosis, Micrognathia, Pierre Robin Sequence,Airway Obstruction,Distraction Osteogenesis

Introduction

Pierre-Robin Syndrome (PRS), also known as Pierre Robin sequence, is a congenital condition that affects the lower jaw and palate. The three main features that characterize PRS arecleft palate (opening in the roof of the mouth), severe underdevelopment of the lower jaw (retrognathia) and glossoptosis (airway obstruction caused by backwards displacement of the tongue base)[1]. This condition occurs in approximately 1 in 8,500 to 14,000 births, and it's equally common in males and females. There is a higher incidence in twins, which may be due to crowding in the uterus, restricting growth of the mandible (lower jaw). PRS may occur in isolation, but it is can be part of an underlying disorder or syndrome. The most common is Stickler Syndrome.The condition is called a sequence because it is believed the underdeveloped mandible begins a sequence of events, which leads to the abnormal displacement of the tongue and subsequent formation of a cleft palate[2].

Inheritance

PRS may also be caused by a genetic disorder. In the case of PRS which is due to a genetic disorder, a hereditary basis has been postulated, but it usually occurs due to a *de-novo* mutation. Specifically, mutations at chromosome 2 (possibly at the GAD1 gene), chromosome 4, chromosome 11 (possibly at the PVRL1 gene), or chromosome 17 (possibly at the SOX9 gene or the KCNJ2 gene) have all been implicated in PRS. Some evidence suggests that genetic dysregulation of the SOX9 gene (which encodes the SOX-9 transcription factor)

and/or the KCNJ2 gene (which encodes the Kir2.1 inward-rectifierpotassiumchannel) impairs the development of certain facial structures, which can lead to PRS. PRS may occur in isolation, but it is often part of an underlying disorder or syndrome. Disorders associated with PRS include Sticklersyndrome, DiGeorgsyndromefetalalcoholsyndrome, TreacherCollinssyndrome, and Patausyndrome[3].

Causes & Risk Factors

An exact cause of PRS is not known. It is believed that any external factor crowds the fetus and interferes with the growth of the lower jaw. Certain neurological conditions, which lead to decreased jaw movement in utero, can also restrict jaw growth. In addition, some studies demonstrate there may be genetic anomalies at chromosomes 2, 11 or 17. Normally, between 9 to 11 weeks of gestation, the tongue moves down and away from the roof of the mouth. This allows space for the sides of the palate to shift to the midline and close[4]. However, in PRS the small mandible keeps the tongue positioned higher in the mouth than normal, thereby interfering with the normal closure of the palate. This typically results in a wide U-shaped cleft of the soft and part of the hard palate.

Symptoms & Types

PRS is commonly characterized by:

An unusual small lower jaw (micrognathia)

Posterior displacement or retraction of the tongue (glossoptosis), which may cause upper airway obstruction

Incomplete closure of the roof of the mouth (cleft palate) is present in the majority of patients, and is commonly U-shaped tongue that is large compared to the jaw

Diagnosis & Tests

There's no specific diagnostic test for PRS. Consulting with a genetic specialist can rule out other problems linked to this syndrome.A diagnosis is made by precise physical examination of the infant, generally right after birth. A CT scan and sleep study may be required. An evaluation must be done with PRS affected infants in a collaborative context to assay the skeletal outcome, outlining air passage hindrance origin, and address minimize hindrance[5]and nourishment issues to augment growth. A multifaceted approach is perfectly suitable for this task, comprises consultants from craniomaxillofacial surgeon, pediatric anesthesia, otolaryngology, pulmonologist, voices therapist, paramedical, and newborn specialist.[8]A genetic specialist should be consulted in case of latent syndrome or hereditary basis. In PRS, tongue retraction[3]may lead to upper air passage hindrance, though these patients can have various other causes for air passage breach due to syndromic reasons. These infants may also have insufficient epiglottis, laryngomalacia, and tracheal stenosis segments, all worsening air passage hindrance.[6 - 8]Airway passage assessment is crucial for PRS treatment decision-making. Significant investigations involve sleep supervision for uncontrolled low oxygen blood concentrations episodes, low oxygen blood concentrations while nourishment, sleep and sound production.[9,10,]Endoscopic examination of velopharynx and endoscopic examination of lungs is helpful complements ascertaining air passage hindrance origin, as there could be more determinants of air passage breach apart from tongue basis, such as in congenital larynx tissue softening above vocal cord, partial tracheal collapse during increased airflow, or other lower larynx hindrances.[11,12]Partial evaluation will involve patient's assessment in various settings and how well this solves upper air passage hindrance.[10,13]

Treatment & Care

A multidisciplinary team approach is highly recommended for proper management of PRS. The goal of treatment is to focus on breathing and feeding and optimizing growth and nutrition despite the predisposition for breathing difficulties. If there is evidence of airway obstruction (snorting breathing, apnea, difficulty taking a breath, or

drops in oxygen), then the infant should be placed in the side or prone position, which helps bring the tongue base forward in many children. In moderate cases, the patient will need to have a tube placed through the nose and into the airways to avoid airway blockage. In severe cases, surgery is needed to prevent a blockage in the upper airway. This is often done by distraction osteogenesis of the lower jaw, which stretches the tissue and grows new bone to make a larger jaw. In rare cases, patients need surgery to make a hole in the windpipe (tracheostomy) because the breathing problems are so severe.Surgery will be needed to repair the cleft palate and this is done between 9 and 12 months of age, depending on the overall health of the child[13,14].

Living & Managing

Children affected with PRS usually reach full development and size. Infants with this condition should NOT be put on their back, to prevent the tongue from falling back into the airway[15,16].

Most children have normal speech after palate repair, but some will require speech therapy or a second operation later on to improve speech.

Feeding must be done very carefully to avoid choking and aspirating liquid into the airway.

Some complications that may occur are:

Breathing difficulties, especially when the child sleeps

Choking episodes

Feeding difficulties

Low blood oxygen and brain damage (due to difficulty breathing)

Conclusion

Pierre robin sequence is a congenital anomaly characterized by micrognathia,cleftpalate and glossotosis at varying levels and presentation. It could be seen as an isolated anomaly or be associated with other congenital conditions or one feature of many syndrome lannelongue and Menard first describes Pierre robin syndrome in 1891 in a report on 2 patients with micrognathia, cleft palate and retroglossotosis. In 1962, Pierre robin published the case of an infant with complete syndrome. Until 1974, the triad was known as Pierre robin syndrome/sequence, however the term syndrome is now reserved for the errors of morphogenesis with simultaneous presence of multiple anomalies caused by 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. Approximately 20 to 40% of cases of Pierre robin sequence are isolated. Isolated Pierre robin sequences affect an estimated 1 in 3500 to 14000 people.A child with PRS may encounter social and emotional challenges unique to this condition as people of the society we have to take care and encourage them in their well-being.

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