Pierre Robin Syndrome

By Jenny Williams

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Overview

Pierre Robin Syndrome, or PRS for short, is a rare developmental disturbance that is distinguished by the presence of glossoptosis, micrognathia, airway obstruction and occasionally, posterior U shaped cleft palate. "In PRS, the micrognathia is so severe that it results in retrognathia, both in the vertical and horizontal facial planes" (Kaufman, Matthew G., et al). In layman's terms, an infant with this condition, would be born with a jaw that is smaller & positioned further back than normal and as a result, their tongue falls posteriorly, jeopardizing their airways and feeding habits. Due to the nature of this condition, perinatal emergencies are common and the mortality rate is as high as 30%.

Etiology

The cause for Pierre Robin Syndrome is unknown but it has been proposed that it may be the result of a mutation in the SOX9 and KCNJ2 genes which are responsible for embryonic mandibular development.

Clinical Presentation

Pierre Robin Syndrome often presents with a smaller than normal sized mandible, posterior positioned tongue and in most but not all cases, cleft palate. Subsequently, this condition also leads to breathing, swallowing and feeding difficulties. Children with PRS commonly have acid-reflux as well. Another interesting presentation is natal teeth where teeth are already erupted in the oral cavity at the time of birth.

Demographic

As of now, Pierre Robin Syndrome does not seem to have any predilections in regards to sex or race. However, it is commonly diagnosed in utero or at the time of birth when clinical signs of the condition are observed.

Biopsy / Histology / Radiographs

There are no histological indications or biopsies that are taken for diagnosis of PRS. However, radiographically, PRS is often detected in utero by way of ultrasonography. The growth of the baby is monitored at multiple stages of the pregnancy and the jaw size at each stage can indicate PRS.

Differential Diagnosis

This condition could possibly be mistaken for Treachers Collins syndrome, Stickler's syndrome, Velocardiofacial syndrome and Marshall's syndrome.

Treatment

There are a plethora of treatment options for PRS as the treatment is based on the clinical presentation and severity for each patient. To treat the obstructed airways, a palatal plate can be applied as well as pharyngeal extensions. It can also be surgically treated through performance of Glossopexy, hyomandibular pexy, subperiosteal release of the mouth floor or tracheotomy. For the resolution of feeding issues, a feeding obturator is also utilized.

Prognosis

Without treatment, the prognosis of Pierrie Robin syndrome is not good as the complications can lead to death from not being able to breathe or eat adequately and aspiration of food into the trachea. Also, in the presence of cleft palate, nasal regurgitation is common which leads to an

infection of the nasopharynx. With treatment, the rate of mortality is significantly lessened and those impacted by the condition are able to live normal lives.

Professional Relevance

As a hygienist it is important to be aware of this condition because our patients may present with it and it's good to have some understanding of what it is. If you're a hygienist in a hospital setting, you would be more likely to see it because it is observed and managed around the time of birth and you may need to take impressions of the infant for construction of a feeding obturator. In this case it's good to know of the condition because it will help you manage the patient better. You will have to be aware of the breathing issues and the need for the patient to be in a supine position. Also, if you don't work in a hospital setting, it is important to know about the condition so that you address the patient correctly. By the time we see the patient they would have already had any necessary surgical treatments performed but still present with the micrognathia. They may be self-conscious about their appearance and therefore we as professionals need to be understanding of their presentations. Lastly, patients with PRS commonly have acid reflux and that is also important to know because of its impact on the enamel.

Citations

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