Pierre-Robin Syndrome

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Overview

Pierre- Robin syndrome (PRS) is a rare congenital condition which affects the head and neck of an infant consisting of a small lower jaw, a tongue that falls back in the throat and difficulty in breathing. This condition was originally described by Dr. Pierre Robin as a form of respiratory obstruction that appeared to be caused by the backward and downward fall of the base of the tongue, a phenotype named glossoptosis. The disease is a chain of developmental malformations, each leading to the next.

Etiology

Exact causes are unknown. It is linked to chromosomal abnormalities near the SOX9 (collagen genes) in the neural crest results in a very short, PRS mandible due to a failure to form Meckel's cartilage.

Clinical Presentation

This condition is characterized by a smaller than normal lower jaw (micronagthia). The hypoplastic mandible provides less volume in the oral cavity and forces the tongue to fit into a smaller space which further serves to aggravate the blockage of the posterior pharynx. Airway obstruction is a result of the abnormal positioning of the tongue (glossoptosis), which serves to occlude the nasal and oral pharynx on inspiration. Blockage of airway passage at the level of the tongue base would result to low blood oxygen concentration, apnea and cyanosis. Child may also have a cleft palate, a posterior U-shaped. Patient is mouth-breather, snores and is using cleft palate as airway. Feeding difficulties are common as infants struggle to breathe during eating. Gastroesophageal reflux and aspiration are common after effect of this process.

Demographic

Highest occurrence rate in white population and lowest in the non-Hispanic black. This is more common in male than female, newborn/infants less than a year old.

Biopsy / Histology / Radiographs

No biopsy is reported for this condition. Histologically, it showed 43.9% to have a connective tissue dysplasia. A patient with dysplasia has developed the RS prenatally due to the abnormal tissue formation in the mandible, larynx and pharynx and will continue having abnormal tissue afterbirth. Calcified tissue are shown in radiographs.

Differential Diagnosis

A wide range of chromosomal anomalies (2, 11 and 17) have been associated with PRS indicating that diverse genetic lesions can trigger abnormal developmental events resulting in similar

phenotype. Conditions that are associated with PRS are Stickler syndrome, Velocardial syndrome (Shprintzen-Goldberg syndrome) and Treacher-Collins syndrome (mandibulo-facial dystosis). Stickler syndrome is characterized by a short mandibular ramus and antegonial notching of the mandibular body. Velocardial syndrome is characterized by a retrognathic mandible and palatal abnormalities. Treacher-Collins syndrome is notable for dysplasia affecting mandible, facial bone hypoplasia, macromastia and high-arching palate.

Treatment

PRS treatment options depend on the condition of the patient. In mild cases, positioning of the child in a side or face-down position to allow the tongue to fall forward and relieve the obstruction. The degree of airway obstruction can be assessed through sleep study (polysomnography) in a hospital setting. If the child has grown enough, nasopharyngeal airway can be used to bring the tongue into a more favorable position. Nasoencopy and bronchoscopy are adjuncts to determining the site of the airway obstruction as there maybe more sources of airway compromise than the tongue itself such as laryngomalacia or subglottic constructions. In severe cases of PRS patients who have abnormal study results, surgical alternatives can be considered such as tongue-lip adhesion and tracheostomy. In tongue-lip adhesion, the undersurface of the tip of the tongue is sutured to the inside of the lower lip to hold in a more forward position. In tracheostomy surgery, a tracheostomy tube effectively bypass the obstruction in the oral pharynx and hypopharynx. When the obstruction is resolved, the tracheostomy tube can be removed. This treatment requires close monitoring. If the tracheostomy tube becomes occluded or dislodged, the patient could have an acute respiratory arrest. Another recently developed technique for treating airway obstruction in PRS is Distraction Osteogenesis. It was popularized by Sidman who has the world's widest experience in treating patients with PRS. In this technique, mandible is cut near the angle of the mandible on both sides. A mechanical device is attached to the mandible approximately 1.5 to 2 mm a day. In 2 to 3 weeks, mandible gradually elongates. When the portions of mandible are separated, new bone is formed. This technique can be performed to a newborn infant to prevent a tracheostomy surgery.

Prognosis

All infants with significant PRS are at risk for sudden death. A 10% mortality rate was documented (0.1 to 5.9 years old). It is high risk when infants sleep in the prone position. Infants with PRS already have compromised airway, and they also typically in the prone positioning. Patients should be treated with a multidisciplinary approach that involves a knowledgeable and experienced medical practitioners capable of providing a comprehensive assessment. Family with these patients should engage in the early stage.

Professional Relevance

There are more consequences as the child grows up and PRS will affect the child's tooth development. Breathing difficulties and nutrition disorder may have been the reason for the enamel defects. Trauma may usually presents to the primary incisors. Tooth anomalies are increasingly considered as an extended phenotype of the palate population. Greater prevalence in the mandible than in the maxilla. There is also a positive correlation between the extent of the palate cleft and the severity of the hypodontia. Partial tooth agenesis or hypodontia is frequently observed in PRS,

a survey was conducted in Norway and Canada. Patient population with isolated RS were surveyed for the presence of hypodontia (excluding third molars). In both of these studies, the dominant pattern of tooth agenesis was the bilateral absence of the mandibular second premolars.

As a dental hygienist, knowledge about this syndrome is important in dental management. Dealing with PRS patient is a challenge. This condition is categorized of children with special needs, received comprehensive dental and orthodontic treatment. They are often exposed to increased of oral diseases, therefore it is crucial to establish a preventive dental home. Recommended preventive instructions should include daily fluoride rinses, fluoride toothpaste, flossing, low carbohydrate diet, periodic professional prophylaxis and fluoride varnish application. The importance of early intervention in children with PRS and multidisciplinary team work from pediatric dentist, oral surgeon, orthodontic specialist and dental hygienist should be strongly considered to give these patients a better quality outcome in life.

Citations

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