Pierre Robin Syndrome

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Abstract- *Pierre Robin sequence is a set of abnormalities affecting the head and face, consisting of a small lower jaw (micrognathia), a tongue that is placed further back than normal (glossoptosis), and blockage (obstruction) of the airways. Most people with Pierre Robin sequence are also born with an opening in the roof of the mouth (a cleft palate).*

Keywords- Pierre Robin sequence, abnormalities, micrognathia, glossoptosis, airways.

I. INTRODUCTION

Pierre Robin syndrome (PRS) also known as Pierre Robin sequence, Pierre Robin malformation, Pierre Robin anomaly or Pierre Robin anomalad. It is a congenital condition of facial abnormalities in humans. PRS is a sequence, i.e. a chain of certain developmental malformations, one entailing the next.

This is condition present at birth, wherein infants are born with micrognathia or unusually small lower jaw, resulting to the tongue that is drawn back into the throat and breathing difficulties. Studies show that the prevalence rate for this condition is 1 in every 8,500 live births, and the male to female ratio is 1:1. In addition, it is possible that Pierre Robin Syndrome can be transmitted through autosomal recessive inheritance.

EPIDEMIOLOGY

The prevalence has been estimated at 1 in 10,000 births, but exact values are hard to know because some that have the symptoms of Pierre-Robin sequence (without any other associated malformation).

ETIOLOGY

Idiopathic Etiology is not knownhowever, there have been various theories formulated to explain its origin.

During the stage of the formation of the bones of the fetus, the tip of the jaw (mandible) becomes 'stuck' in the point where each of the collar bones (clavicle) meet (the sternum), effectively preventing the jaw bones from growing. It is thought that, at about 12 to 14 weeks gestation, when the fetus begins to move, the movement of the head causes the jaw to "pop out' of the collar bones. From this time on, the jaw of the fetus grows as it would normally, with the result that, when born, the jaw of the baby is much smaller (micrognathia) than it would have been with normal development, although it does continue to grow at a normal rate until the child reaches maturity.

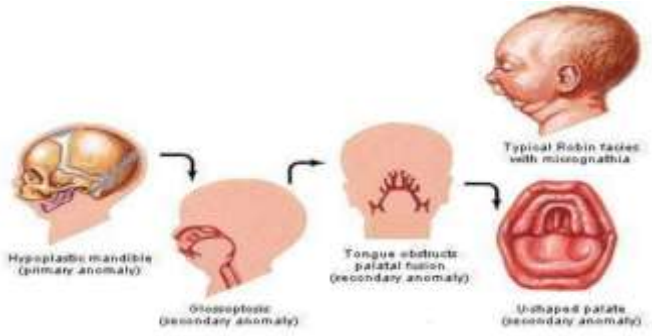
Genetic Dysregulation Pierre Robin sequence may be caused by genetic anomalies at chromosomes. Recent studies have indicated that genetic dysregulation of SOX9 gene prevents the SOX9 protein from properly controlling the development of facial structures. Autosomal recessive inheritance is possible. An X-linked variant has been reported involving cardiac malformations and clubfeet.

MAIN FEATURES

- cleft palate
- retrognathia (abnormal positioning of the jaw or mandible) and
- glossoptosis (airway obstruction caused by backwards displacement of the tongue base)
- a small lower jaw (micrognathia),

PATHOGENESIS

Mechanical Theory This is the most accepted theory explains that the cleft palate, the first event that happens in patients with Pierre Robin syndrome, occurs between the seventh and eleventh week of gestation. During these months, the tongue is high in the oral cavity, forming a cleft in the palate to prevent the closure of the palatal shelves. Theory thus explains the inverted U-shaped cleft and the absence of the associated cleft lip.



PRS may occur in isolation, but it is often part of an underlying disorder or syndrome. Most common is Stickler Syndrome. Other disorders include Velocardiofacial syndrome, Fetal Alcohol Syndrome and Treacher Collins Syndrome.

Neurological Maturation Theory: This theory supports the idea that there has been a delay neurological maturation as noted in the electromyography testing on the muscles of the tongue, pharyngeal pillars, and the palate as well as a lag in the conduction of the hypoglossal nerve, which is one of the nerves that innervate the tongue.

Rhombencephalic Dysneurulation Theory: The disorganization of the motor regulation of the hindbrain or the rhombencephalus causes major problems of ontogenesis

II. SIGNS AND SYMPTOMS

PRS is characterized by

- an unusually small mandible (micrognathia),
- posterior displacement or retraction of the tongue (glossoptosis), and
- 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.

The following are the symptoms of Pierre Robin Syndrome Micrograthia

- Smaller than normal jaw size

Characteristics:

- Inferior dental arch is retracted behind the superior arch
- The mandible has smaller body, obtuse genial angle, and condyles are posteriorly located Glossoptosis

Retraction of the tongue occurs in 70-85 percent of case

- Cleft soft palate
- High-arched palate
- Larger tongue compared to the jaw
- Natal teeth – teeth already appear by the time the baby is born
- Otitis media – ear infection
- Small opening in the roof of the mouth, which might cause choking

The occurrence of both small jaw size and the retraction of the tongue will usually cause severe respiratory and feeding difficulty in the newborn

III. DIAGNOSIS

- The syndrome is diagnosed shortly after birth due to characteristic facies.
- The infant usually has respiratory difficulty, especially when supine.
- Tongue which tends to ball up at the back of the mouth and fall back towards the throat (glossoptosis))
- Small lower jaw (micrognathia)
- The cleft palate is U-shaped(horse shoe)
- Feeding difficulties

IV. MANAGEMENT

The **goals of treatment** in infants with Robin sequence focus upon breathing and feeding and optimizing growth and nutrition despite the predisposition for breathing difficulties.

Conservative Management

FOR RESPIRATORY SYMPTOMS

Airway Obstruction (snorty breathing, apnea, difficulty taking a breath, or drops in oxygen), then

- the infant should be placed in the side lying or prone position,
- Oral airway placement,
- laryngeal mask or intubation in severe refractory cases

FOR FEEDING DIFFICULTIES

- Upright feeding techniques, modification of the nipple for bottle feeding, temporary use of nasogastric or orogastric feeding tube, and the placement of a gastrostomy.
- Palatal plates such as the pre- epiglottic baton plate, which have a velar extension, pulls the base of the tongue forward
- This can be helpful in the relief of airway obstruction, and it also facilitates the swallowing mechanism during feeds
- include upright positioning on a wedge (a tucker sling may be needed if the baby is in the prone position), small and frequent feedings (to minimize vomiting), and/or pharmacotherapy (such as proton pump inhibitors).

SURGICAL MANGAMENT

According to the severity of the condition i.e infants with pronounced micrognathia may experience severe respiratory distress or failure to thrive due to feeding difficulties.

- Tracheostomy : life saving in severe airway obstruction
- Glossopexy: (Routledge Procedure) ¬ but should be released before start of denitation and speech development
- Mandibular lengthening i.e Distraction Surgery: ¬gradual distraction may be used for severe mandibular hypoplasia that causes obstructive apnea.

DIFFERNITIAL DIAGNOSIS

- Childhood Sleep Apnea
- DiGeorge Syndrome (DGS)
- Fetal Alcohol Syndrome
- MandibulofacialDysostosis (Treacher Collins Syndrome)
- CHARGE Syndrome

PROGNOSIS

- In spite of airway obsstruction problems and feeding difficulties , the prognosis in PRS is mostly good.
- With proper management PRS babies can grow to healthy normal adult life.

COMPLICATIONS

These complications can occur:

- Breathing difficulties, especially when the child sleeps
- Choking episodes
- Congestive heart failure
- Death
- Feeding difficulties
- Low blood oxygen and brain damage (due to difficulty breathing)
- Type of high blood pressure called pulmonary hypertension

PREVENTION

There is no known prevention. Treatment may reduce breathing problems and choking.

REFERENCES

[1] Jakobsen, LP; Knudsen, MA; Lespinasse, J; GarcíaAyuso, C; Ramos, C; Fryns, JP; Bugge, M; Tommerup, N (March 2006). "The genetic basis of the Pierre Robin Sequence.". The Cleft Palate-Craniofacial Journal. 43 (2):155 9. PMID 16526920. doi:10.1597/ 05-008 .1.

[2] "Pierre Robin syndrome" at Dorland's Medical Dictionary

[3] R, S; A, MP (May 2013). "Role of SOX9 in the Etiology of Pierre-Robin Syndrome.". Iranian journal of basic medical sciences. 16 (5): 700–4. PMC 3700045. PMID 23826492.

[4] Jakobsen, LP; Ullmann, R; Christensen, SB; Jensen, KE; Mølsted, K; Henriksen, KF; Hansen, C; Knudsen, MA; Larsen, LA; Tommerup, N; Tümer, Z (June 2007). "Pierre Robin sequence may be caused by dysregulation of SOX9 and KCNJ2.". Journal of MedicalGenetics. 44 (6):386. PMC 2740883 . PMID 17551083. doi: 10.1136/ jmg.2006.046177.

[5] Jaiswal, SK; Sukla, KK; Gupta, V; Rai, AK (December 2014). "Overlap of Patau and Pierre Robin syndromes along with abnormal metabolism: an interesting case study." (PDF). Journal of Genetics. 93 (3): 865-8. PMID25572249. doi:10.1007/s12041-014-0452-2.

[6] Van den Elzen AP, Semmekrot BA, Bongers EM, Huygen PL, Marres HA (January 2001). "Diagnosis and treatment of the Pierre Robin sequence: results of a retrospective clinical study and review of the literature". Eur. J. Pediatr. 160 (1): 47–53. PMID 11195018. doi:10.1007/s004310000646.

[7] "Isolated Pierre Robin syndrome". Orphanet. Retrieved 17 May 2015.

[8] Pierre Robin. La glossoptose. Son diagnostic, ses consequences, son traitement. Bulletin de

l'Académienationale de médecine, Paris, 1923, 89: 37. Journal de médecine de Paris, 1923, 43: 235-237.

[9] "Noel Rosa 100 anos". O Página Cultural. Retrieved March 1, 2015.