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Care of child with Pierre robin sequence and its nursing management

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Abstract
Pierre Robin Sequence (PRS) is a rare congenital birth defect wherein one malformation leads to a chain of events causing the formation of other anomalies. The first abnormality that is developed is Micrognathia leading to glossoptosis and, ultimately to airway obstruction and/or a cleft palate in an affected patient. It has a higher incidence among twins. Although the exact cause of the defect is unknown but it can be caused by mutations in various chromosomes. There are two types of this defect: isolated/Non Syndromic and Syndromic Pierre Robin Sequence (PRS). There are numerous other differential diagnoses with near to similar clinical manifestations; thus, a genetic evaluation should be considered for infants to identify a specific syndromic diagnosis and various other diagnostic tests are further required to rule out the functional and structural defects in the patient. Management of the defect is patient oriented, multi-phased and surgery is performed to solve the functional problems often associated with airway impairment of the patients.

Keywords: PRS, Pierre robin sequence, pierre robin syndrome, robin pierre syndrome, robin sequence.

Introduction
Pierre Robin Sequence (PRS) is also known as Pierre Robin Syndrome or Pierre Robin malformation or even Robin Syndrome. It is a rare congenital birth defect characterized by a small lower jaw (micrognathia) and pushed back tongue (glossoptosis) and upper airway obstruction. Some infants may also have an abnormal opening in the roof of the mouth (cleft palate) [1]. The clinical features of this defect can be present either as an isolated sequence or as a part of a genetic syndrome. Though the cardinal triad features of cleft palate, micrognathia, and airway obstruction was first identified by St Hilaire in 1822, then by Fairbain in 1846, and followed by Shukowsky in 1911 [2]. It was a French stomatologist, Pierre Robin who first published a case of an infant with glossoptosis and explained its anatomical relationship with micrognathia and associated airway complications in 1923 [3]. He also discussed the management by "liberation of the oral pharynx" with a prosthetic device, pulling the jaw and tongue forward [4, 5] and also reported growth failure and death are caused by the respiratory complications that occur due to micrognathia and glossoptosis [6].

Risk Group: Although Pierre Robin sequence (PRS) affects both males and females equally, with an estimated prevalence rate of about 1 in 8,500-14,000 individuals; still there is a higher incidence among twins [7].

Etiology: The exact cause of Pierre Robin Sequence (PRS) is unknown. Although there are multiple contributing factors, PRS is believed to be caused by mutations on various chromosomes [8].

Types
There are two types of Pierre Robin Sequence (PRS)

1. **Isolated PRS/Non Syndromic PRS:** When Pierre Robin Sequence (PRS) occurs by itself.
2. **Syndromic PRS:** When Pierre Robin Sequence (PRS) occurs in multiple anomaly disorders.

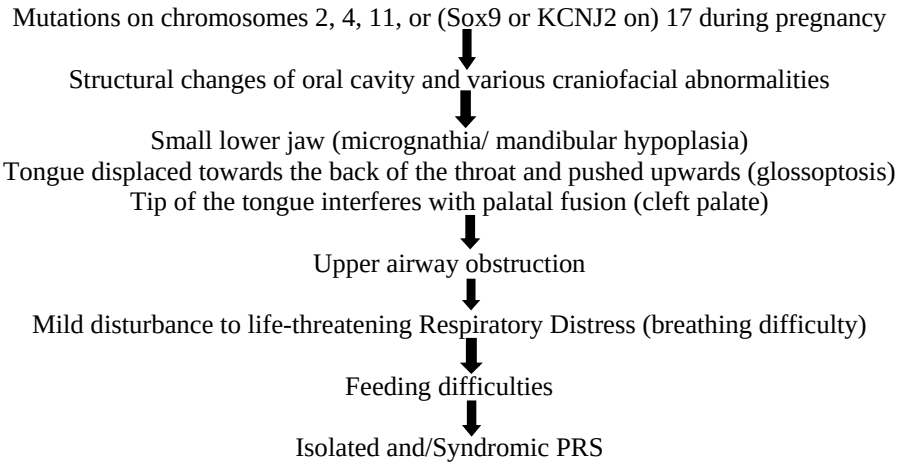
In Isolated/ Non Syndromic PRS, DNA near a gene called Sox9 is affected. The Sox9 gene is responsible for the production of the Sox9 protein, which is crucial for skeletal development. In affected individuals, there are mutations in this DNA that alters the Sox9's activity. When these areas get damaged, the Sox9 gene's activity is reduced, leading to less normal Sox9 protein production. This is believed to cause craniofacial abnormalities which are characteristically associated with Pierre Robin sequence (PRS) [7]. Jakobsen *et al*. compared data in several databases and listed the genes that might lead to the cause of Pierre Robin sequence (PRS), including GAD67 on 2q31, PVRL1 on 11q23-q24 and Sox9 on 17q24.3-q25.1 [9, 10] Melkoniemi *et al*. also detected disease-associated mutations in *COL11A1* and *COL11A2* genes in some patients with nonsyndromicPierre Robin sequence (PRS) [10, 11].

Jakobsen *et al*. screened 10 unrelated patients affected with PRS for Sox9 and KCNJ2 mutations and revealed that nonsyndromicPierre Robin Sequence (PRS) may be caused by both Sox9 and KCNJ2 dysregulation [10, 12]. It is seen that isolated Pierre Robin Sequence (PRS) are believed to arise sporadically, or through new genetic changes, rather than being inherited. Syndromic Pierre Robin sequence (PRS) on the other hand is inherited following the same genetic pattern.

Alternatively, syndromic Pierre Robin Sequence (PRS) accounts for 60% of PRS [13]. There have been 34 syndromes associated with syndromic PRS, the most common being Stickler syndrome (accounts 20-25%) [14, 10] followed by Velocardiofacial syndrome (accounts 15%) [15]. Treachery Collins syndrome (*mandibulofacial dysostosis*), Nager syndrome, *Spondyloepiphyseal* dysplasia congenital (SEC), and other recognized syndromes account for the rest of the syndromic PRS cases.

Pathophysiology

Pierre Robin syndrome (PRS) is now named as a sequence because one malformation leads to a chain of events causing the formation of other anomalies. In Pierre Robin sequence (PRS), *micrognathia* is the first abnormality that leads to *glossoptosis* and, ultimately, *airway obstruction* and/or a cleft palate.



There are few common theories regarding hypo plastic mandibular growth, which are as follows

1. Mechanical Theory: Around the 7th week of gestation, the mandibular growth becomes abnormal leading to the abnormal trajectory of growth of the tongue which then blocks the closure of the palatal cleft in the 11th week of gestation. This glossoptosis ultimately leads to upper airway obstruction [8, 16].

2. Neurological Maturation Theory: There is a delay in the neuromuscular development of the tongue, which does not stimulate the mandible to grow normally or palatal shelves to fuse properly [8, 16].

3. Mandible Compression Theory: The external forces cause the fetal head to become flexed, pushing the mandible against the chest, making it unable to grow properly. The causes can be either multifetal gestation, uterine anomalies, or oligohydramnios. Though the tongue continues to grow normally but is ultimately displaced backwards, leading to obstruction of the upper airway. The tip of the tongue then obstructs the fusion of the palatal shelves, leading to cleft palate [8, 16].

Clinical Manifestations

Micrognathia: There is a shorter mandibular length and a large mandibular angle.

Glossoptosis: There is displacement of the base of the tongue to the pharynx. There is a wide range of severity depending on the amount of displacement and therefore associated respiratory distress.

Airway Obstruction: Clinical signs include abnormal breathing sounds, increased respiratory accessory muscle use, desaturations, difficulty feeding/swallowing, reflux, and aspiration. Long term manifestations may include reduced weight gain, difficulty speaking, neurological deficits, and ultimately pulmonary hypertension and corn pulmonale. Airway obstruction with Pierre Robin Syndrome (PRS), if very severe or not properly managed, may lead to hypoxia, corn pulmonale, failure to thrive, and cerebral impairment. Syndromic cases and Robin complexes are more severe than nonsyndromic PRS and have worse prognoses. Mortality may be as high as 30%. [17] Neonates with Pierre Robin Syndrome (PRS) should be carefully monitored because a significant airway obstruction may develop during the first

1-4 weeks of life. 'Obstructive sleep apnea' can occur during the night. In this condition the breathing temporarily halts and restarts because of periodic blockage of the airways.

Cleft Palate: U-shaped cleft palate is the most common followed by V-shaped type, both types affects breathing and speech development.

Feeding Difficulties: It occurs due to abnormal oral cavity. Depending on the severity, there can be choking (due to aspiration) or 'failure to thrive', acid (gastro esophageal) reflux in affected children.

Other Possible Manifestations: Cardiovascular and lung conditions, such as heart murmurs, pulmonary hypertension and pulmonary stenosis. Musculoskeletal system anomalies, including those in the arms, legs, feet, and vertebral column. Repeated otitis media (80% of patients) and ocular defects (10% to 30% of patients) [7]. Natal teeth are among other frequent findings.

Differential Diagnosis

Beckwith-Wiedemann Syndrome, CHARGE Syndrome, Childhood Sleep Apnea, DiGeorge Syndrome, Fetal Alcohol Syndrome, Mandibulofacial Dysostosis (Treacher Collins Syndrome), Multiple Births, Pectus Excavatum, Pediatric Cleft Lip and Palate, Stridor, Velocardiofacial Syndrome. Thus, a genetic evaluation should be considered for infants with Pierre Robin Syndrome (PRS) to identify a specific syndromic diagnosis and provide recommendations for genetic testing.

Diagnostic Tests

- Three-dimensional ultrasound to identify micrognathia at third trimester (prenatal).
- Antenatal sonographic visualization of glossoptosis (prenatal).
- Physical examination to quantify the small or posteriorly displaced jaw and other craniofacial abnormalities.
- Ophthalmologic screening along with molecular testing (including Stickler syndrome-associated collagen gene analysis and fluorescence in situ hybridization for 22q11.2 deletion syndrome) as to detect retinal detachment and blindness in children [22].
- Nasoendoscopy and Nasopharyngoscopy to detect upper airway obstruction at the tongue base along with vocal cord mobility (caused by glossoptosis).
- Bronchoscopy to evaluate subglottic structures, including the trachea and bronchi.
- Genetic Evaluation (fluorescence in situ hybridization [FISH]) to identify 22q deletion, mutation in Treacle [*TCOF1*] gene.
- Cephalograms (Lateral Radiographs) to identify the facial profile, mandibular length, and distance between the upper and lower alveoli.
- A jaw-thrust maneuver performed under anesthesia with direct endoscopic visualization to predict dynamic airway change with mandibular advancement.
- Bone radiographs to assess for other suspected syndromes.
- Continuous oxygen saturations to detect desaturations with feeding, sleep, and different positions.
- Polysomnography to establish the severity of obstructive apneic events and the need and timing of intervention.
- Feeding specialist and/ Speech pathologist to detect feeding problems.

Management

Management of Pierre Robin Sequence (PRS) is multi-phased and individualized. Surgery is only performed to solve the functional problems often associated with airway impairment of the patients.

Breathing difficulties

In many cases, airway difficulties may be mild at birth but can progress during the first 4-8 weeks of life. As a newborn's respiratory needs are relatively small as compared to an infant who grows on a faster rate thus the requirements increase and making the obstruction more severe. Subsequently infants with a Non-syndromic Pierre Robin Sequence (PRS) will often outgrow this type of obstruction. One should never neglect the need to identify the type of airway obstruction and understand its mechanism for a correct management and treatment.

- Prone position (to prevent the tongue from falling back towards the throat resulting in a larger airway passage).
- Oral airways (for larger airway passage).
- 'Nasopharyngeal airway' insertion into the nose (to keep the airways open).
- Continuous positive airway pressure or endotracheal intubation, mechanical ventilation to manage severe respiratory distress.

In cases where the infant can only maintain CO_2 levels above 50, a surgical procedure is mandatory. The following three surgical procedures are used to treat PRS [18, 19]

- **Tongue-lip adhesion/glossopexy** (the tongue is sutured to the lower lip, thereby pulling the tongue forward and providing a larger airway. Later, presumably after the child has demonstrated catchup growth, this bond between the tongue and lip is separated)
- **Tracheostomy** (it effectively bypasses the obstruction for better air passage and once the obstruction is resolved, the tracheostomy tube can be removed)
- **Distraction osteogenesis** of the mandible for severe isolated PRS cases and for syndromic PRS cases in which mandibular catchup growth does not occur. The mandible is cut near the angle of the mandible on both sides. A mechanical device distracts the two portions of the mandible approximately 1.5-2 mm a day. As the portions of the mandible are separated, new bone is formed, and the mandible gradually elongates over a period of 2-3 weeks.

Feeding-Related Difficulties

- Keeping the child's head more elevated and using specially designed cleft bottles and nipples may be used.
- If severe, a gavage/feeding/ gastrostomy tube may be inserted temporarily for proper weight gain.

Cleft Palate

Palatal closure surgery is conducted between 12-18 months of age. Surgery to improve the jaw's appearance is rarely

require because the small lower jaw most often grows to a more normal size by the age of 18 months. If a child already has a tracheostomy tube in-situ, the palate repair can be performed at any time. In some cases with a micro gnathic jaw and a normal tongue size, an infant might be using his/her cleft palate as an airway passage. Closing the cleft may compromise the airway function; therefore, a multidisciplinary team of specialists must carefully evaluate the timing of cleft palate closure.

Symptomatic and supportive treatment

- For impaired speech, speech therapy can be provided by a speech pathologist.
- For ear and hearing-related issues, otolaryngologists and audiologists can provide individualized therapies and associated care.
- For recurrent ear infections, surgically placed drainage tubes and appropriate pharmacological management can be opted.
- For avoiding crowding of the teeth and for proper tooth alignment a team of orthodontists, maxillofacial surgeons, and dentists can work together.
- For ocular abnormalities, ophthalmologist may be consulted.
- Genetic counseling shall be of beneficial for patients and their families.

Complications

The complications of Pierre Robin Sequence (PRS) are mainly due to airway obstruction. Short term complications include desaturations, difficulty in feeding, and aspiration. Long term complications attributes to hypoxic injury and inability to feed properly. This may also include cerebral impairment, pulmonary hypertension, cor pulmonale, and failure to thrive. Procedural complications relating to PRS are discussed in the treatment/management section. Early detection of PRS can help prevent long term complications of airway obstruction and hypoxemia.

Nursing Management

Management of a patient diagnosed with Pierre Robin Sequence requires an interprofessional team efforts where in the nurses plays a pivotal role in maintaining the balance between the management of various specialists including maternal-fetal medicine (MFM) specialists, family planning specialists, neonatologists, plastic surgeons, otolaryngologists, anesthesiologists, oral maxillofacial surgeons, dentists, dieticians, speech pathologists, and geneticists [20].

Pre Natal Nursing Management Aligning the counselling sessions for the parents

- Geneticist and a maternal-fetal medicine specialistto diagnose the defect.
- Neonatologists to discuss the immediate assessment and care of the newborn in the first weeks to months of life, including airway management and typical feeding/growth difficulties associated with the defect.
- Otolaryngologists and oral maxillofacial surgeons to provide information regarding potential surgical options and related complications.
- Dieticians to provide necessary information about various ways to solve issues related to feeding difficulties and failure to thrive.
- Speech pathologists to discuss about the training needed for consequent speech and swallowing problems.

Initial Neonatal Nursing Management

- Admit to the NICU following birth and when stabilized transfer to post ICU/high dependency unit for ongoing management.
- Clinical examination of the baby to evaluate for syndromic association and refer to genetics for inpatient review if concerns.
- Frequently maintaining prone position, if clinically significant airway obstruction despite prone position, consider inserting Nasopharyngeal Airway (NPA) or supporting airway with CPAP/NIV.
- Preparing for Intubation in case of severe cases of upper airway obstruction if indicated/needed.
- Regular Cardiorespiratory monitoring
- Conducting complete microarray (parental consent required) when advised by the physician.
- Arranging the consultations for clinical evaluation for the severity of micrognathia.
- Discuss with the parents the need for clinical 3D facial photo and obtain consent and arrange for it.
- Assess the suck feeding along with the feeding team.
- Prepare consultation schedules with the ENT team for a Flexible Nasopharyngeal Endoscopy (FNE) in the first week of life.
- Assist the sleep study evaluation during the day time at 7-10 days of age to diagnose sleep study in half supine and half prone sleep position.
- Arrange appointment schedules with Plastic Surgeons for reviewing the extend of retrognathia/micrognathia.

Nursing Care of Naso Pharyngeal Tube (in-situ)

- Suction tube 2-3 hourly for the first 48-96 hours, then whenever indicated or needed.
- Observe and document at least each shift, condition of the skin around nares, and under taping.
- Change tape if soiled with milk or secretions and remove the tape with adhesive remover.

Nursing care for feeding

- Place the hand between the infant's shoulders and the neck to support the infant during feeding. The infant should 'sit' on the thigh. And his/her head/neck should not be extended but the spine should be straight OR Position the infant on the lap or on a pillow on the lap in a side lying position. The infant's neck and spine should be in a natural straight alignment and hips should be flexed at 90 degrees.
- Use the Haberman Feeding bottle/MAM Squeezy Bottle with teat (s)/Pigeon squeezy bottle, place the nipple in the side of the infant's mouth and gently squeeze the bottle to allow sufficient milk to flow in the infant's mouth for them to swallow without choking. Co-ordinate squeeze, sucking and swallowing.
- Constant monitoring (cardiac, SaO2 and direct vision) throughout the feed.
- Closely monitor the baby's cues throughout the feed and provide breaks between the feed if stress is identified.

- Excessive drooling/ frothing of milk happens if the teat is under the tongue. Remove the teat and place it on top of the tongue.
- Burping on regular intervals and whenever required (Keep the infant in an upright forward leaning position to allow the infant's jaw to fall forward, thereby preventing airway obstruction during the burping).
- If the feed is prolonged (30 minutes or more) provide the feed via NGT to prevent infant's exhaustion.

Nursing care for palate closure and surgical correction

- Essential newborn/infant care to be provided (warmth, immunization, infection prevention, hygienic care, etc).
- Prepare the newborn/infant for the respective surgery/constructive surgeries as per the condition.
- Close observation and monitoring of vital signs, bleeding from the surgical sites, oral secretions, vomiting and crying, especially after the concerned surgery/surgeries.
- Place the newborn/infant on the abdomen after the surgery.
- Mummify/swadle the newborn for emotional touch
- Perform mouth care and care of the suture line with normal saline, H_2O_2 and antiseptic ointments as prescribed.
- Keep the newborn/infant dry, well fed and comfortable to prevent crying. Use Logan Bow id need for suture protection in the mouth.
- Avoid sucking, crying and talking for palatal repair healing
- Provide nasogastric feeding initially and later replace it with small frequent liquid feeds.
- IV therapy to be administered as prescribed for nutritional and hydration support.
- Analgesics, antibiotics and other relevant pharmacological management to be administered as prescribed as per the clinical manifestations exhibited.
- Provide play and diversional therapies for the newborn/infant.
- Arrange suture removal after 5-14days or as per the repair healing.
- Align the consultation schedules with the other specialist for the newborn/infant as needed.

Discharge Planning: Aim to discharge by 3-6 weeks of age.

Infant must exhibit

- No bedside desaturations.
- No to minimal upper airway obstruction on sleep evaluation.
- Safe feeding on assessment by the feeding team
- Adequate weight gain; 150-200 grams/week.

Newborn/infant must be reviewed by ENT CNS, Feeding Team, Plastic Surgeons, and Respiratory Team prior to discharge.

Parent / Caregiver must be

- Confident in feeding the infant.
- Confident with the basic care/positioning of their infant.
- Confident in feeding the newborn/infant.
- Able to identify the danger signs in newborn/infant at home.
- Educated for safe infant sleeping and travelling with the newborn/infant after discharge.

Conclusion

Pierre Robin Syndrome (PRS) is a rare defect which is characterized by the formation of one malformation which then leads to a chain of other structural anomalies. It is clinically presented by the presence of micrognathia followed by glossoptosis and ultimately to airway obstruction and/or a cleft palate along with various other associated structural and functional health problems.

A genetic evaluation is a must to diagnose Pierre Robin syndrome (PRS) from other differential diagnoses with near to similar signs and symptoms. Management of Pierre Robin sequence (PRS) requires multiple interventions and is individualized as per the patient's functional problems.

References

1. Hsieh ST, Woo AS. Pierre Robin Sequence. Clin Plast Surg. 2019;46(2):249-259.
2. St-Hilaire H, Buchbinder D. Maxillofacial pathology and management of Pierre Robin sequence. Otolaryngol Clin North Am. 2000;33(6):1241-1256
3. Gangopadhyay N, Mendonca DA, Woo AS. Pierre Robin Sequence. Semin Plast Surg. 2012;26(2):76-82.
4. Robin P. A drop of the base of the tongue considered as a new cause of nasopharyngeal respiratory impairment [In French]. Bull Acad Natl Med (Paris). 1923;89:37-41
5. Robin P. A fall of the base of the tongue considered as a new cause of nasopharyngeal respiratory impairment: Pierre Robin Sequence, a translation-1923. Plast Reconstr Surg. 1994;93(6):1301-1303
6. Robin P. Glossoptosis due to atresia and hypotrophy of the mandible. Am J Dis Child. 1934;48:541-547.
7. Internet
   Pierre Robin Sequence. NORD Web site. Pierre Robin Sequence - Symptoms, Causes, Treatment | NORD (rarediseases.org) Accessed January 08, 2024.
8. Diana Baxter, Anthony L. Shanks. Pierre Robin Syndrome. NIH National Library of Medicine. 2023 August 7; Bookshelf ID: NBK562213PMID: 32965884.
9. Jakobsen LP, Knudsen MA, Lespinasse J, Garcia Ayusa C, Ramos C, Fryns JP, *et al*. The genetic basis of the Pierre Robin Sequence. Cleft PalateCraniofac J. 2006;43(2):155-9.
10. Internet: Pierre Robin Sequence. MEDSCAPE Web site. Pierre Robin Sequence: Background, Pathophysiology, Etiology (medscape.com) Accessed January 08 2024.
11. Melkoniemi M, Koillinen H, Mannikko M, Warman ML, PihlajammaT, Kaariainen H, *et al*. Collagen XI sequence variations in non syndromic cleft palate, Robin sequence and micrognathia. Eur J Hum Genet. 2003;11(3):265-70.
12. Jakobsen LP, Ullmann R, Christensen SB, Jensen KE, Molsted K, Henriksen KF, *et al.* Pierre Robin Sequence may be caused by dysregulation of Sox9 and KCNJ2. J Med Genet. 2007;44(6):381-6.
13. Izumi K, Konczal LL, Mitchell AL, Jones MC. Underlying genetic diagnosis of Pierre Robin Sequence: Retrospective chart review at two children's hospitals

and a systematic literature review. J Pediatr. 2012;160(4):645-650.e2.
14. Karempelis P, Hagen M, Morrell N, Roby BB. Associated syndromes in patients with Pierre Robin Sequence. Int. J Pediatric Otorhinolaryngology. 2020;131:109842.
15. Shprintzen RJ, Goldberg RB, Young D, Wolford L. The velo-cardio-facial syndrome: a clinical and genetic analysis. Paediatric. 1981;67(2):167-72.
16. Giudice A, Barone S, Belhous K, Morice A, Soupre V, Bennardo F, *et al*. Pierre Robin sequence: A comprehensive narrative review of the literature over time. J Stomatology Oral Maxillofacial Surg. 2018;119(5):419-428.
17. Jones KL, Jones MC, Campo DM. Robin sequence. Smith's recognizable patterns of human malformation. 8th Edition. Philadelphia: Elsevier; c2022. p. 334-7.
18. Internet: Pierre Robin Syndrome. Cleveland Clinic Website. Pierre Robin Syndrome (Sequence): Facts, Surgery, causes, symptoms & what it is (clevelandclinic.org) Accessed January 08 2024.
19. Hong P. A clinical narrative review of mandibular distraction ontogenesis in neonates with Pierre Robin sequence. Int. J Pediatric Otorhinolaryngology 2011;75(8):985-91.
20. Internet: Pierre Robin Sequence - Information Documents for Parents. Pierre Robin Europe. Pierre Robin Sequence - Information Document for Parents (pierrerobineurope.com) Accessed January 08 2024.