Pierre Robin Sequence

Pierre Robin Sequence (PRS) is a sequence of facial abnormalities caused by underdevelopment of the mandible leading to secondary defects.

Pathophysiology

Sequence mechanism:

- **1) Mandibular hypoplasia/micrognathia –** small jaw results in tongue failing to descend
- **2) Glossoptosis –** tongue is pushed superior and posterior into the pharynx
- **3) Airway obstruction** due to the position of the tongue

- In up to 85% of PRS cases, the palatal shelves are unable to fuse, resulting in a **U-shaped cleft palate**
- Pierre Robin is a ***sequence*** – because one primary anomaly (small mandible) causes a chain of secondary defects

Etiology

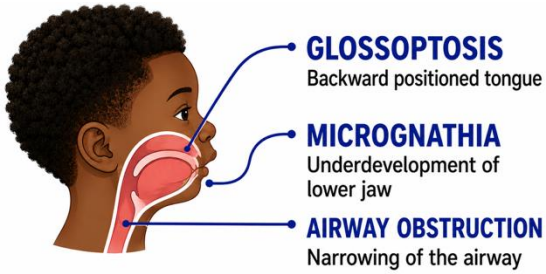
- **Primary mandibular hypoplasia** during weeks 7-11 of gestation
- PRS may be **isolated** or **syndromic**, with approximately 20-40% of cases being isolated
- May result from **mechanical constraints** (i.e. oligohydramnios) or **genetic mutations** (i.e. SOX9/COL2A1)

Diagnosis

Clinical diagnosis based on triad: **micrognathia, glossoptosis,** and **airway obstruction** +/- cleft palate

Investigations:

- Prenatal US: signs of micrognathia
- Airway assessment: flexible laryngoscopy
- SLP for feeding assessment
- Polysomnography

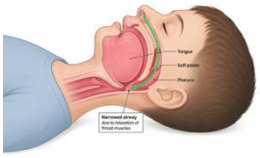


Cleft palate

Presentation

Neonatal symptoms

- Respiratory distress, cyanosis, stridor (worse supine, improves when prone)
- Feeding difficulty and growth faltering
- Sleep disordered breathing, snoring



Nasopharyngeal airway

Management

- **Airway support:** prone positioning, nasopharyngeal airway, CPAP, intubation
- **Feeding support:** Special bottles (Haberman feeder), nasogastric or gastrostomy feeding if necessary
- **Definitive/Surgical Management:** Mandibular distraction osteogenesis **(MDO),** tongue-lip adhesion **(TLA**), tracheostomy for severe obstruction, eventual cleft palate repair (~1 year of age)
- ** Note: most isolated cases improve as mandible grows with age, without requiring surgical intervention*
- **Multidisciplinary care:** Plastics, ENT, NICU, OMFS, SLP, Genetics, Nutrition

July 2026

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