# PIERRE ROBIN SEQUENCE

## Otherwise known as?

Pierre Robin sequence is also known by the following names:

1. Pierre Robin syndrome
2. Robin sequence
3. Pierre Robin anomaly
4. Pierre Robin malformation

## Signs & Symptoms

Pierre Robin sequence is a condition that involves a combination of craniofacial differences. The primary features include micrognathia (a small jaw), cleft palate, and airway obstruction. The severity of symptoms can vary from mild to severe, but the following are the main signs and symptoms associated with Pierre Robin sequence:

### 1. Micrognathia (Small Jaw)

- Receding chin or small jaw: The most characteristic feature of Pierre Robin sequence is a small lower jaw. This results in a receding chin, which can make it difficult for a baby to feed properly.
- Underdeveloped jaw can cause the tongue to fall backward, leading to airway obstruction.

### 2. Cleft Palate

- Cleft palate: Many infants with Pierre Robin sequence are born with a cleft palate, which is a gap or opening in the roof of the mouth. This can lead to:
  - Difficulty with feeding, as it can interfere with sucking and swallowing.
  - Problems with speech development as the child grows.
  - Increased risk of ear infections due to problems with drainage from the middle ear.

### 3. Airway Obstruction

- Breathing difficulties: Due to the small jaw (micrognathia), the tongue may fall back into the throat and block the airway, causing respiratory distress, especially in the early days of life.
  - Babies may struggle to breathe and may require medical assistance such as nasal airways (also known as nasal airway adjuncts or nasal passages devices, which are medical tools or techniques used to help maintain an open airway in the nose), positioning (keeping the baby on their stomach or side to help with breathing), or, in severe cases, tracheostomy (a surgically created hole in the windpipe).

*(continued overleaf)*

  - Obstructive sleep apnoea is common in some children with Pierre Robin sequence due to airway problems.

### 4. Feeding Problems

- Due to the combination of a small jaw and cleft palate, babies may have difficulty breastfeeding or bottle-feeding. They may not be able to create a good seal to suck effectively, leading to:
  - Difficulty gaining weight.
  - Feeding times may be longer or more exhausting.
  - Some babies may require special feeding techniques, such as using a cleft palate bottle or special nipples.

### 5. Speech and Language Delays

- Speech delays are common because of the cleft palate, which can impact the ability to produce certain sounds.
- Children with Pierre Robin sequence may experience challenges with articulation and nasal speech due to the gap in the roof of the mouth.
- Early speech therapy can help improve these skills over time.

### 6. Ear Difficulties

- Children with Pierre Robin sequence are more prone to middle ear infections (otitis media), which can lead to hearing problems if left untreated.
- The differing palate can affect the Eustachian tubes (which help drain the middle ear), increasing the risk of fluid build-up and infections.

### 7. Feeding and Developmental Delays

- Some children with Pierre Robin sequence may have global developmental delays, including delays in motor skills (like crawling and walking) due to their physical and feeding challenges.
- Delayed growth or difficulty maintaining weight might also occur in some cases, especially if feeding problems are severe.

## Causes (*the why*)

Pierre Robin sequence is often a congenital condition, meaning it is present at birth. The exact cause of the sequence can vary, but it is generally caused by genetic factors or impaired development of the jaw and palate during pregnancy.

*(continued overleaf)*

# PIERRE ROBIN SEQUENCE

### 1. Isolated Pierre Robin Sequence (non-syndromic)

In many cases, Pierre Robin sequence occurs sporadically without being associated with any other health problems or syndromes. This is known as isolated or non-syndromic Pierre Robin sequence. The cause in these cases is usually unknown, but it is thought to result from an irregularity during foetal development that affects the development of the lower jaw (mandible).

- Early developmental disturbance: During the early stages of pregnancy, there is a problem with the development of the jaw, which leads to micrognathia (a small or underdeveloped jaw). This can cause the tongue to be positioned differently, blocking the formation of the palate and leading to a cleft palate.
- This irregular development might not always have a clear cause, and it can happen randomly in some pregnancies.

### 2. Genetic Causes (Syndromic Pierre Robin Sequence)

Pierre Robin sequence can also occur as part of a genetic syndrome, in which other birth conditions or health issues are present. These genetic conditions can have an autosomal dominant or autosomal recessive inheritance pattern, meaning they can be inherited from one or both parents.

### 3. Environmental Factors

Although genetic factors are the primary cause, some environmental factors may contribute to the development of Pierre Robin sequence.

### 4. Unknown Causes

In some cases, no specific cause can be identified, and Pierre Robin sequence may occur due to random genetic mutations or early developmental disruptions that are not fully understood.

## Testing & Diagnosis

The diagnosis of Pierre Robin sequence is based on clinical evaluation, which includes a thorough assessment of the baby's physical features, medical history, and symptoms. If Pierre Robin sequence is suspected, further genetic testing and imaging may be done to confirm the diagnosis and rule out associated conditions.

### 1. Clinical Evaluation

The first step in diagnosing Pierre Robin sequence is a physical examination by a doctor. During the clinical evaluation, the following features are typically observed:

*(continued overleaf)*

- Micrognathia: A small or recessed lower jaw, which causes the chin to appear underdeveloped.
- Cleft palate: A gap or opening in the roof of the mouth, which may be complete or incomplete.
- Breathing difficulties: The small jaw can cause the tongue to obstruct the airway, leading to respiratory distress or apnoea (temporary cessation of breathing), especially in newborns.
- Feeding problems: Difficulty with breastfeeding or bottle-feeding due to the cleft palate and small jaw, leading to poor weight gain.

If these features are present, the doctor will likely diagnose Pierre Robin sequence based on these clinical signs.

### 2. Genetic Testing

Genetic testing is an important step, particularly if the doctor suspects the Pierre Robin sequence may be part of a syndrome (e.g., Stickler syndrome or 22q11.2 deletion syndrome). Genetic testing can help identify any underlying genetic causes or mutations.

Tests That May Be Ordered Include:

- Chromosome microarray analysis: This test looks for deletions or duplications of chromosomes that might be responsible for genetic syndromes, such as 22q11.2 deletion syndrome.
- DNA sequencing: If a specific syndrome is suspected (like Stickler syndrome), the doctor may recommend sequencing of specific genes associated with that condition.
- Whole exome sequencing (WES): This is a more comprehensive genetic test that sequences all protein-coding genes and may help identify rarer or less understood genetic causes.

### 3. Imaging Studies

Imaging studies can be helpful for assessing the degree of craniofacial differences and ruling out associated problems.

- Ultrasound: In some cases, a prenatal ultrasound may detect irregular craniofacial features before birth, such as a small jaw or cleft palate. This can help identify the need for postnatal follow-up.
- X-rays or CT scans: After birth, imaging may be performed to evaluate the structure of the jaw, skull, and palate. This helps in planning surgical interventions if necessary. CT scans may be used to assess the skull and airway in more detail.

*(continued overleaf)*

- MRI: An MRI may be done in some cases to assess the brain and detect any associated problems such as hydrocephalus (fluid build-up in the brain), particularly if there are concerns about neurological issues.

### 4. Evaluating Associated Conditions

Since Pierre Robin sequence can sometimes be part of a broader syndrome, additional tests may be performed to evaluate for any associated conditions, such as:

- Ophthalmological exams: If Stickler syndrome is suspected, an eye exam can check for issues like retinal problems or vision impairment.
- Heart evaluation: Some syndromes associated with Pierre Robin sequence, such as 22q11.2 deletion syndrome, can involve heart problems. A cardiologist may perform an echocardiogram to assess heart function.
- Hearing tests: Hearing loss is common in some genetic syndromes, so an audiology exam may be done to assess hearing function in newborns and children with Pierre Robin sequence.

### 5. Multidisciplinary Approach

Because Pierre Robin sequence can involve several body systems (e.g., craniofacial, respiratory, and feeding systems), the diagnosis often involves a multidisciplinary team of specialists, including:

- Paediatrician or geneticist: To assess and interpret clinical features and genetic tests.
- Craniofacial surgeon or plastic surgeon: To evaluate the need for surgical intervention to address the cleft palate or jaw differences.
- Speech pathologist: To assess feeding and speech development.
- Paediatric pulmonologist: To evaluate respiratory issues and breathing problems.
- Otolaryngologist (ENT specialist): To assess ear, nose, and throat function, particularly in relation to feeding and hearing.
- Audiologist: To test hearing function and detect potential hearing loss.

## Treatment

The treatment of Pierre Robin sequence focuses on managing the airway, feeding difficulties, and any associated craniofacial differences. The treatment plan can vary depending on the severity of the condition, the presence of a cleft palate, and whether Pierre Robin sequence is isolated or part of a broader syndrome. A multidisciplinary team of specialists typically works together to address the various aspects of the condition.

*(continued overleaf)*

# PEIRRE ROBIN SEQUENCE

## 1. Airway Management

Because micrognathia (a small jaw) often causes the tongue to block the airway, leading to respiratory distress or apnoea, airway management is the most immediate concern, especially in newborns.

Methods for Airway Management:

- Positioning: Keeping the infant in a prone position (on their stomach) or side-lying position can help prevent the tongue from blocking the airway. This is often done in the first few months of life when airway obstruction is most common.
- Nasopharyngeal airway: A nasal airway may be inserted to help keep the airway open, especially if the baby experiences severe breathing difficulties.
- CPAP or BiPAP: For babies with significant respiratory distress, continuous positive airway pressure (CPAP) or bilevel positive airway pressure (BiPAP) may be used to provide positive pressure to keep the airway open during breathing.
- Tracheostomy: In rare and severe cases where breathing difficulties cannot be controlled, a tracheostomy (a surgical opening in the windpipe) may be required to bypass the blocked airway.
- Feeding tube: In cases of severe respiratory distress, a feeding tube (nasogastric or gastrostomy tube) may be used to ensure the baby gets adequate nutrition while the airway issues are being managed.

## 2. Feeding Support

Due to the cleft palate and small jaw, infants with Pierre Robin sequence often have trouble sucking and swallowing, which can lead to poor weight gain and dehydration.

Feeding interventions may include:

- Specialized bottles and nipples: There are special cleft palate bottles and nipple designs that help create a better seal and allow the baby to feed more easily.
- Feeding therapy: A speech-language pathologist or occupational therapist with experience in feeding can help teach parents techniques for safe feeding, ensuring the baby gets enough milk and avoids aspiration (food entering the lungs).
- Tube feeding: In cases where feeding is particularly difficult or if the infant is not gaining weight, a feeding tube (nasogastric tube or gastrostomy tube) may be placed temporarily to provide nourishment directly to the stomach.

*(continued overleaf)*

# PEIRRE ROBIN SEQUENCE

### 3. Surgical Treatment

Surgical interventions are often required to address cleft palate, jaw differences, and improve breathing over time.

Common surgeries include:

- Cleft Palate Repair:
    - Surgical repair of the cleft palate is typically performed around 9-12 months of age. This surgery closes the gap in the roof of the mouth, improving feeding and speech development.
    - Early repair of the cleft palate helps reduce the risk of ear infections and speech problems later in life.
- Tracheostomy (if necessary):
    - If the child has severe airway obstruction that cannot be managed with less invasive treatments, a tracheostomy may be performed to allow the child to breathe more easily. This is more common in severe cases.
- Ear tubes:
    - Due to the risk of middle ear infections, children with Pierre Robin sequence may need ear tube placement to help prevent fluid build-up in the ears, which can lead to hearing loss

### 4. Speech and Language Therapy

- Speech therapy is crucial for children with a cleft palate or who have had difficulty with feeding, as they may experience delays in speech development.
- Early intervention in the form of speech therapy can help children with Pierre Robin sequence develop clearer speech and improve articulation.
- Palate therapy: In some cases, children may benefit from a special appliance or device to help improve palate function.

### 5. Monitoring and Ongoing Care

- Regular follow-up visits: Children with Pierre Robin sequence often require regular follow-up appointments with a multidisciplinary team, including paediatricians, craniofacial surgeons, speech therapists, audiologists, and geneticists.
- Hearing assessments: Due to the risk of middle ear infections, hearing tests should be conducted regularly to monitor for any hearing loss.
- Dental and orthodontic care: As children grow, they may need orthodontic treatment to address any jaw or dental issues that arise from the cleft palate or jaw differences.

*(continued overleaf)*

# PEIRRE ROBIN SEQUENCE

## 6. Genetic Counselling and Family Support

- Genetic counselling: If Pierre Robin sequence is part of a genetic syndrome, genetic counselling can help families understand the potential for recurrence in future pregnancies and provide guidance on managing the syndrome.
- Psychosocial support: Because Pierre Robin sequence can involve complex medical care and surgeries, families may benefit from psychosocial support or connecting with support groups for advice and emotional support.

**Did you know?**

In Australia, approximately 19 babies per 100,000 births are diagnosed with Pierre Robin sequence. *

Pierre Robin sequence was first described by French physician Pierre Robin in 1923. He identified a pattern of craniofacial differences, including a small jaw (micrognathia), a cleft palate, and respiratory difficulties caused by the tongue's position.

While Robin initially recognized the sequence, it wasn't until later that it was understood as part of a broader spectrum of developmental issues, sometimes occurring in association with other genetic syndromes.

## References:

*Junaid, M., Slack-Smith, L., Wong, K., Bourke, J., Baynam, G., Calache, H., & Leonard, H. (2022). Epidemiology of Rare Craniofacial Anomalies: Retrospective Western Australian Population Data Linkage Study. The Journal of Pediatrics, 241, 162-172.e169 https://doi.org/10.1016/j.jpeds.2021.09.060