CONDITION

Pierre robin sequence

Pierre robin sequence is a birth condition involving a small lower jaw, a backward-positioned tongue, and possible airway blockage.

WHAT IT IS
Linked differences in jaw, tongue, and airway development

BODY SYSTEM
Face, mouth, upper airway, and feeding

MAIN SYMPTOMS
Small lower jaw, noisy breathing, feeding difficulty, poor growth

WHAT CAUSES IT
Restricted jaw growth, sometimes linked to genetic conditions

HOW IT IS DIAGNOSED
Clinical examination, with airway and feeding assessments

MAIN TREATMENT
Airway support, feeding support, and sometimes surgery

CAN IT BE CURED?
Sometimes - breathing often improves as the jaw grows

IS IT AN EMERGENCY?
Sometimes - blue color, severe breathing difficulty, or breathing pauses

LONG-TERM OUTLOOK
Often good with early airway, feeding, hearing, and speech care

Overview

Pierre robin sequence, also called Robin sequence, is a group of facial and airway differences present at birth. It is called a "sequence" because one developmental difference leads to another.

The lower jaw is smaller or set farther back than usual. This is called micrognathia . The small jaw leaves less room for the tongue, which can fall backward toward the throat. This is called glossoptosis . The tongue may partly block the airway, especially during sleep or feeding.

Many babies also have a cleft palate, meaning there is an opening in the roof of the mouth. The cleft is often wide and U-shaped. It develops because the tongue's position prevents the two sides of the palate from joining before birth. A cleft palate is common in Pierre robin sequence, but it is not required for the diagnosis.

The main early concerns are breathing, safe feeding, and growth. The degree of airway blockage can range from mild to severe. Some babies need only close monitoring and feeding support. Others need an airway device or surgery.

Pierre robin sequence may occur by itself. It may also be part of a genetic condition that affects other parts of the body. This distinction can influence follow-up and long-term expectations.

Symptoms

The features are usually noticed at birth or soon afterward. Breathing problems may be more obvious when a baby lies flat, sleeps, cries, or feeds.

Possible signs include:

Some babies have obstructive sleep apnoea , in which the upper airway repeatedly narrows or closes during sleep. Signs can include snoring, restless sleep, chest pulling inward, pauses in breathing, sweating, or frequent waking. Babies do not always appear distressed even when their oxygen level is falling, so medical assessment is important.

Feeding difficulty may come from both the cleft palate and the blocked airway. A baby may have trouble creating suction, tire quickly, or need frequent pauses to breathe. Coughing or choking during feeds can suggest that milk is entering the airway, known as aspiration.

- A small or backward-positioned lower jaw
- A tongue that sits farther back in the mouth
- Noisy, high-pitched, or effortful breathing
- Pauses in breathing or changes in skin color
- A cleft palate
- Difficulty sucking, swallowing, or coordinating breathing with feeding
- Milk coming through the nose
- Long or tiring feeds
- Slow weight gain

Causes and risk factors

Pierre robin sequence begins during early development before birth. The lower jaw does not

grow forward as expected. The tongue then stays high and backward in the mouth, where it may affect both the airway and the developing palate.

Why jaw growth is restricted is not always known. The sequence can be isolated, meaning no broader condition is identified. In other babies, it is associated with a genetic or chromosome difference.

Associated conditions include Stickler syndrome, 22q11.2 deletion syndrome, and Treacher Collins syndrome. These conditions have different features and inheritance patterns. Having Pierre robin sequence does not mean that a baby necessarily has one of them.

Some genetic changes are inherited from a parent. Others happen for the first time in the child. The condition is not caused by anything a parent did during pregnancy.

A clinical genetics assessment may be suggested when there are other physical differences, developmental concerns, hearing or eye findings, or a family history of similar features.

Diagnosis

Diagnosis is mainly based on an examination after birth. The care team looks at the size and position of the jaw, the tongue, the palate, and the baby's breathing.

Assessment does not stop with the appearance of the face. A baby who seems comfortable while awake may still have airway blockage during sleep or feeding. Monitoring may include oxygen measurements and observation in different positions.

A thin flexible camera passed through the nose, called nasendoscopy , may be used to see where the airway is narrowing. A sleep study may record breathing effort, airflow, oxygen, and carbon dioxide during sleep. These tests help measure severity and guide treatment rather than establish the sequence by themselves.

Feeding specialists may observe a feed. If swallowing safety is uncertain, an imaging test may show whether milk enters the airway.

Hearing tests are important because a cleft palate can affect pressure and fluid drainage in the middle ear. Genetic testing may be offered when an associated condition is suspected. Testing can sometimes identify a cause, but a normal result does not rule out every genetic condition.

Pierre robin sequence can occasionally be suspected on an ultrasound before birth. Prenatal imaging cannot always show how much breathing or feeding support a baby will need after delivery.

Treatment and management

Treatment focuses first on keeping the airway open and making feeding safe. Care is often coordinated by a craniofacial or cleft team. This may include specialists in newborn care, ear, nose and throat care, breathing, surgery, feeding, speech, hearing, dentistry, and genetics.

Some babies breathe adequately with careful positioning while they are being medically monitored. Because infant sleep position also affects the risk of sleep-related death, families need a specific sleep plan from their specialist team. Positioning used in hospital should not be copied at home without clear instructions.

If positioning is not enough, a soft tube may be placed through the nose to hold the airway open. Some babies use continuous positive airway pressure, or CPAP, which gently pushes air through a mask. Oxygen alone may not correct an obstruction and does not replace proper airway assessment.

Feeding support may include pacing, upright or side-lying techniques, and bottles designed for babies with a cleft palate. Expressed breast milk or formula can be given with these systems. If feeding by mouth is unsafe or uses too much energy, a tube through the nose may provide some or all nutrition. Longer-term tube feeding through the stomach is sometimes needed.

Surgery may be considered when significant obstruction continues despite less invasive support. Options include moving the lower jaw forward gradually, attaching the tongue temporarily closer to the lower lip, or creating an airway opening in the neck. The choice reflects the location and severity of the blockage, the baby's anatomy, and any associated condition.

Cleft palate repair is usually planned separately, once breathing is stable and the surgical team considers the timing appropriate. Later care may include hearing treatment, speech therapy, dental care, and orthodontics.

Complications

Untreated airway obstruction can lead to low oxygen levels, excess carbon dioxide, disrupted sleep, and strain on the heart and lungs. Early monitoring and treatment reduce these risks.

Feeding problems can cause dehydration, slow growth, or aspiration. Poor growth may also make surgery and recovery more difficult.

Fluid can collect behind the eardrum because the palate muscles do not open the middle-ear drainage tubes normally. Repeated fluid or infections may contribute to hearing loss. Reduced hearing can affect speech development if it is not recognised and managed.

A repaired cleft palate may still affect speech. Some children have air escape through the nose while speaking or need further assessment of palate movement.

Dental crowding and bite differences can develop as the jaws and teeth grow. Some people continue to have a smaller lower jaw or sleep-disordered breathing later in childhood or adulthood.

The chance of other complications is influenced by whether Pierre robin sequence is isolated or part of a wider condition.

Prevention

Pierre robin sequence usually cannot be prevented. It develops before birth, and there is no established action during pregnancy that reliably prevents it.

If a genetic cause is found, a genetics professional can explain the chance of it occurring in another pregnancy. That chance is different for each genetic condition and family. When no cause is identified, an exact recurrence risk may not be possible to give.

Early recognition does not prevent the sequence, but it can help prevent complications from airway blockage, unsafe swallowing, poor growth, hearing loss, and delayed speech support.

Living with it and what to expect

The first weeks and months usually focus on breathing, feeding, and weight gain. Families may need to learn how to use special bottles, an airway tube, feeding equipment, or monitoring devices. Follow-up is often more frequent during this stage.

As the lower jaw grows forward, the tongue may have more room and airway obstruction may improve. This commonly happens during infancy and early childhood, but the timing and amount of improvement cannot be predicted for an individual child. Babies with an associated syndrome may follow a different course.

Feeding often becomes easier as breathing improves and the palate is repaired. Some children still need feeding therapy because of swallowing difficulty, oral sensitivity, or limited experience eating by mouth.

Cleft, hearing, speech, dental, and jaw care can continue through childhood and adolescence. Hearing checks are needed even when a child seems to respond normally to sound. Speech may develop typically with treatment, but some children need therapy or another procedure.

Children with isolated Pierre robin sequence often have typical learning and development. The

sequence itself does not mean a child has an intellectual disability. Development may be affected if there has been prolonged low oxygen, severe feeding difficulty, or an associated genetic condition.

Many people take part in ordinary school, work, exercise, and family life. Some continue to need care for their bite, jaw position, hearing, speech, or sleep-related breathing as they grow.

When to get medical help

Call emergency services if a baby or child:

Seek urgent medical assessment for worsening noisy breathing, new breathing pauses, repeated choking during feeds, or signs of dehydration. Signs of dehydration include fewer wet nappies, a very dry mouth, unusual sleepiness, or difficulty taking feeds.

Contact the regular care team promptly if feeds are taking much longer, weight gain has slowed, an airway tube will not stay in place, or symptoms are changing. Fever, ear pain, ear discharge, or reduced response to sound also need routine medical review.

Planned follow-up remains important even when breathing appears better. Airway obstruction can be most noticeable during sleep, and hearing loss from middle-ear fluid may have few obvious signs.

- Turns blue, grey, or unusually pale around the lips or face
- Stops breathing, has repeated breathing pauses, or cannot be woken normally
- Has severe chest pulling, gasping, or obvious difficulty getting air
- Becomes limp or suddenly much less responsive
- Chokes and cannot breathe or make sounds

Common questions

Is Pierre robin sequence the same as a cleft palate?

No. Pierre robin sequence involves a small lower jaw and a tongue positioned toward the throat, with resulting airway concerns. A cleft palate is common, but some babies with the sequence do not have one. Cleft palate also occurs without Pierre robin sequence.

Will my baby outgrow it?

Jaw growth often improves the amount of space behind the tongue, so breathing may become easier over time. This is not guaranteed, and a cleft palate does not close through growth.

Associated genetic conditions, airway anatomy, and the severity of early obstruction all affect the course.

Can a baby with Pierre robin sequence sleep on their back?

Safe sleep needs individual planning because lying flat may worsen obstruction in some babies, while non-back sleep positions carry their own risks. Follow the written plan from the baby's airway or craniofacial team. Do not use sleep positioners or change a prescribed plan without medical advice.

Can my baby breastfeed?

Some babies can feed at the breast, especially if there is no cleft and breathing is stable. A cleft palate often prevents the suction needed to remove enough milk. Expressed breast milk can be offered using a specialist bottle when appropriate. A feeding assessment can help identify the safest method.

Is Pierre robin sequence genetic?

Sometimes. It may occur by itself without an identified genetic cause, or as part of an inherited or newly occurring genetic condition. Genetic assessment is more likely to be recommended when there are other health or physical findings.

Does it affect intelligence?

Isolated Pierre robin sequence does not usually affect intelligence directly. Developmental outcomes may differ when the sequence is part of another condition or when severe breathing and nutrition problems have occurred.

Sources

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