

Head: Pierre Robin Sequence

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Copy: In 1923, a physician named Pierre Robin described a newborn child with an abnormally small lower jaw (mandible), large tongue and breathing problems. Today, Pierre Robin Sequence (PRS) is a condition of facial difference characterized by severe underdevelopment of the lower jaw (retrognathia), a downward or backward-positioned tongue (glossoptosis) and usually a cleft palate (opening in the roof of the mouth).

A sequence, not a syndrome

The disorder is called a sequence and not a syndrome because the underdeveloped lower jaw begins a sequence of events, which lead to the abnormal displacement of the tongue and subsequent formation of a cleft palate.

During the normal developmental process, between nine to eleven weeks during gestation, the tongue moves down and away from the roof of the mouth, allowing space for the sides of the palate to shift to the midline and close. In PRS, the small lower jaw keeps the tongue positioned higher in the mouth than normal, thereby interfering with the normal closure of the palate. This typically results in a wide U-shaped cleft of the soft and part of the hard palate.

The overall incidence of Pierre Robin Sequence is low, approximately one in 8,500 to 14,000 births, and equally common in males and females. The diagnosis of PRS is made by examining the infant and not by special diagnostic tests. The exact cause of PRS is not known. External factors, which crowd the fetus and interfere with the growth of the lower jaw, may contribute to PRS.

There is a higher incidence of PRS in twins, which may be due to crowding in the uterus, thereby restricting growth of the lower jaw. Certain neurological conditions, which lead to decreased jaw movement in utero, can also restrict jaw growth. If an individual has PRS because of the influence of external factors while in utero, his or her risk of passing on the condition is minimal, because the genetic information governing jaw and palate development has not been altered.

Possible genetic basis

Some studies demonstrate there may be a genetic basis for PRS. PRS can be seen in other syndromes including Stickler and Velocardiofacial Syndrome.

Stickler is the most common syndrome associated with PRS, occurring in 10 to 30 percent of cases. Stickler Syndrome is a genetic malfunction in the tissue that connects bones, heart, eyes and ears. This disorder is associated with problems with vision, hearing, bones and joints, the heart and facial formation, including cleft palate.

Velocardiofacial Syndrome is associated with PRS in approximately 10 percent of cases. The most common features are cleft palate, heart defects,

characteristic facial appearance, minor learning problems, and speech and feeding problems.

In isolated cases where PRS is not associated with any other syndromes, the risk of a parent having another child with PRS is three to five percent. The risk of an adult with isolated PRS having a child with cleft palate is three to five percent. Genetic testing may be offered if a genetic syndrome is suspected. Chromosome analysis and testing for chromosome deletion (FISH for deletion of 22q11) can be performed.

Feeding difficulties

Nearly every newborn with PRS will experience some degree of feeding difficulty. This is because of a combination of factors, including the size of the lower jaw, position of the tongue and the cleft of the palate. Babies with minor degrees of PRS can learn to feed with specially adapted nipples and bottles such as the Haberman Feeder, the Meade Johnson Cleft Palate Nurer or a regular nipple with a larger opening. A feeding consultant can often help parents chose the right nipple/bottle combination through a course of trial and error.

For infants with more sever PRS, the risk of aspiration during feeding can be high. In addition, the baby can struggle to move milk to the back of the throat and swallow while working on breathing, and significant calories can be lost.

To prevent aspiration during feeding and to allow the child to gain weight appropriately, a feeding tube (nasogastric tube or NG tube) may be inserted into the nose and down into the stomach. This is a safe procedure that will ensure the infant obtains the needed calories to gain weight appropriately. This is a temporary solution that can be used for up to a month.

Children who require long-term feeding assistance may need a gastric tube surgically inserted through the abdominal wall into the stomach. Many children with PRS outgrow their feeding problems by one to two years of age, when the mandible grows more sufficiently.

Respiratory problems

Breathing problems can be common in infants with PRS. A normal-sized lower jaw helps to anchor the tongue in a more forward position. Because of the small, recessed jaw found in children with PRS, the tongue tends to fall backwards and block off the throat and obstruct breathing when the infant is placed on his/her back.

This is of great concern during sleep, when the tongue is more relaxed and prone to fall back into the throat. The majority of babies respond well to positioning on the stomach which helps pull the tongue forward during sleep. Other infants may require nasal tubes or surgery to pull or push the tongue forward.

At our hospital, we institute a trial of positioning the infant on his/her stomach. If this works to relieve the respiratory obstruction, infants can safely be sent home with an apnea monitor.

If stomach positioning does not work at relieving the respiratory obstruction, a nasopharyngeal airway may be passed through the nose into the upper airway to help with breathing. A nasal airway can be used for a short period of time. Occasionally a sleep study will be done prior to discharge from the hospital to insure the child is safe to be sent home without risk of significant apnea.

For those infants with more severe respiratory obstruction, surgical procedures may be required to improve breathing. For children whose breathing obstruction is not relieved by prone positioning, we generally recommend a surgical procedure called a tongue-lip adhesion.

A tongue-lip adhesion is a safe procedure which temporarily sutures the tip of the tongue to the inside of the lower lip thereby pulling the tongue forward and out of the back of the throat. The tongue-lip adhesion stays in place for eight to ten months, until the lower jaw has grown enough to pull the tongue forward on its own.

Mandibular distraction

Some hospitals will perform a procedure called mandibular distraction (figure 1) if positioning on the stomach or a tongue lip adhesion fails to relieve the respiratory obstruction. Mandibular distraction is a procedure involving surgically cutting the lower jaw and placing either an internal (in the mouth) or external (through the skin) device which can be slowly adjusted to lengthen the jaw and theoretically pull the tongue out of the back of the throat.

Unfortunately, none of these procedures works all of the time, and a small number of children with PRS and severe respiratory obstruction may require a tracheostomy to help with breathing. Most children with isolated PRS experience enough jaw growth during the first one to two years of life to allow for eventual removal of the tracheostomy.

Children with PRS and other associated syndromes such as Stickler or Velocardiofacial may have a small lower jaw for life. For any child with PRS, it is important to have surgical procedures performed at a hospital where there are anesthesiologists familiar with the anesthetic difficulties in children with a small lower jaw.

Cleft palate repair

The cleft palate is usually repaired when the infant is between nine and twelve months old, depending on the health of the child. In PRS, surgery may be postponed a month or two if the lower jaw and mouth are very small. Palate surgery takes about two or three hours, with the infant staying in the hospital one or two nights.

A child with PRS may spend his/her first night after palate surgery in the pediatric intensive care unit to monitor for signs of airway obstruction. A cleft palate can result in abnormalities with the middle ear, leading to persistent fluid, which is a primary cause of repeat ear infections. Hearing loss can be a consequence of repeat ear infections and persistent middle ear fluid. Tubes can be inserted into the ear at the time of palate repair to alleviate fluid build-up and restore hearing.

Most children have normal speech after palate repair, but some will require speech therapy or a second operation to improve speech later on. Because of the complexity of Pierre Robin Sequence and clefts of the palate, PRS should be diagnosed and treated by an experienced team of experts recognized by the American Cleft Palate-Craniofacial Association.

Figure 1: Mandibular distraction

[If you do not have any more pictures, you don't need to use "Figure 1:".]