a guide to understanding

pierre robin sequence

a publication of children's craniofacial association

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This parent's guide to Pierre Robin Sequence is designed to answer questions that are frequently asked by parents of a child with Pierre Robin Sequence. It is intended to provide a clearer understanding of the condition for patients, parents and others.

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what is pierre robin sequence?

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), respiratory obstruction, and usually a cleft palate (opening in the roof of the mouth).

Normally, between nine to eleven weeks of gestation, the tongue moves down and away from the roof of the mouth. This allows space for the sides of the palate to shift to the midline and close. However, 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.

why is pierre robin called a sequence and not a syndrome?

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

what causes pierre robin sequence?

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. Certain neurological conditions which lead to decreased jaw movement in utero, can also restrict jaw growth. In addition, some studies demonstrate there may be a genetic basis for PRS.

how is PRS diagnosed?

The diagnosis of PRS is made by examining the infant and not by special diagnostic tests.

what are the chances of my baby having PRS?

The overall incidence of Pierre Robin Sequence is low, approximately one in 8,500 to 14,000 births. PRS is equally common in males and females. 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.

will my child pass PRS on to his/her children?

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.

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 also 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.

is PRS seen in other syndromes?

PRS can be seen in other syndromes including Stickler and Velocardiofacial syndromes.

Stickler syndrome is the most common syndrome associated with PRS. It occurs in 10 to 30 percent of cases. Stickler syndrome is a genetic malfunction in the tissue that connects bones, heart, eyes and ears. Someone with Stickler syndrome may have 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 of this syndrome are cleft palate, heart defects, characteristic facial appearance, minor learning problems, and speech and feeding problems.

when is the cleft palate repaired?

The cleft palate is usually repaired when an 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 in order to watch for signs of airway obstruction. A cleft palate can result in abnormalities with the middle ear, leading to persistent fluid. This is a primary cause of repeat ear infections. Repeat ear infections and persistent middle ear fluid may result in hearing loss. 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 later on to improve speech. 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.

what other problems might we expect and how will they be treated?

two problems you might expect to deal with in Pierre Robin Sequence are feeding difficulties and respiratory problems.

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 Nurser, or a regular nipple with a larger opening. A feeding consultant can often help parents choose the right nipple/bottle combination through a course of trial and error.

For infants with more severe PRS, the risk of aspiration (inhaling fluid into the lungs) during feeding can be high. In addition, the baby may struggle to move milk to the back of the throat and swallow, while trying to breath. In this case significant calories for weight gain 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 and is a temporary solution that can be used 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 when the mandible grows more sufficiently. This usually occurs by one to two years of age.

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 when the child is placed on his/her back. When the tongue falls backwards it blocks off the throat and obstructs breathing.

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.

Some hospitals may try 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 (a monitor that alerts the parents when breathing is interrupted).

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. This insures 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, a tongue-lip adhesion may be recommended.

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.

In cases where positioning on the stomach or a tongue lip adhesion fails to relieve the respiratory obstruction, some surgeons will perform a procedure called mandibular distraction. (figure 1) 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. This device 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 work all of the time, and a small number of children with PRS and severe respiratory obstruction may require a tracheostomy. A tracheostomy is a surgically created

opening through the neck, into the trachea (breathing tube) to allow the passage of air 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.

how can children's craniofacial association (cca) benefit my family?

CCA understands that when one family member has a craniofacial condition, each person in the family is affected. We provide programs and services designed to address these needs. A detailed list of CCA's programs and services may be found on our website at www.ccakids.org or call us at 800.535.3643.

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