

a guide to understanding
**pierre robin
sequence**



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empowering and giving hope to individuals and families affected by facial differences

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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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This booklet is intended for information purposes only. It is not a recommendation for treatment. Decisions for treatment should be based on mutual agreement with the craniofacial team. Possible complications should be discussed with the physician prior to and throughout treatment.

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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), an upward and/or backward-positioned tongue (glossoptosis), airway obstruction, 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 can result 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 position of the tongue and subsequent airway obstruction.

what are the chances of my baby having PRS?

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

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.

what is the likelihood of having another child with prs, or my child passing it on to his/her children?

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.

Most infants born with PRS had acquired it randomly. So in general, the risk of having another child with PRS is low. The risk is similar to the risk of having another child with a cleft palate, three to five percent.

Some infants born with PRS have a genetic cause that may run in families. Stickler syndrome is the most common. The risk of having another child with PRS in this situation is higher but the severity may be different between 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.

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 can also 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, or if the infant's airway is still obstructed. 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.

Children with a cleft palate almost all have problems draining the fluid from behind the eardrum, leading to persistent fluid. This may lead to repeated ear infections or the fluid may only cause hearing loss. Careful evaluation of the ears is important part of multidisciplinary care of children with cleft palate. 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 may be at an increased risk of needing surgery to improve their speech. Because of the complexity of Pierre Robin Sequence and clefts of the palate, children with PRS should be evaluated and managed 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, they may have difficulty balancing breathing and feeding. Some may even have trouble with aspiration (inhaling fluid into the lungs) during feeding. This makes breathing even more challenging. The combination of feeding problems with burning more calories while struggling to breathe makes it important to monitor all babies with PRS carefully for weight gain.

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 can be a temporary solution for children with feeding and weight gain difficulties.

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. Many 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.

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 ensures the child is safe to be sent home without risk of significant apnea.

For children whose breathing obstruction is not relieved by prone positioning, a surgery to improve the breathing may be recommended. The three main surgeries to improve the airway are (1) tongue-lip adhesion, (2) mandible (jaw) distraction, or (3) tracheostomy. Different centers have a different preference to these treatments.

A tongue-lip adhesion is a 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. Some centers prefer to address the small mandible rather than performing the tongue-lip adhesion and others use it when tongue-lip adhesion fails to relieve the airway obstruction. Mandible (or jaw) distraction is a procedure involving surgically cutting the lower jaw and slowly making the jaw longer. This is done with either an internal

device (embedded under the tissue) or an external device (most of the device is on the outside of the skin). Surgeons use this device to slowly lengthen and sometimes change the shape of the jaw. This pulls the tongue forward and makes more room for breathing.

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, with a breathing tube placed in this opening. This bypasses the obstruction at the tongue and allows children to breathe comfortably.

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. Monitoring the lower jaw for growth and the airway for signs of obstruction, as a child with PRS ages, is important.

Any surgery in infancy can be scary for parents. You should feel comfortable with your child's team and they should be able to answer your questions. The airway in children with PRS can be challenging to manage, so it is important to have surgical procedures performed at a hospital where the anesthesiologists are familiar with the 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. We believe it is helpful to hear the stories of other parents with a child that has PRS. It can also be helpful to share your experiences or concerns with others who are experiencing a similar medical journey. CCA can match you with families who are willing to network with you. If you contact us, we can help you find people in your region that are associated with CCA and people across the country who you may connect with via phone, email, and social media. Your local craniofacial team may also have a network of parents and/or support groups. These resources can help during stressful times but be careful about medical advice from those who haven't received medical training. 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.