What is Pierre Robin Syndrome?

Pierre Robin Syndrome is a congenital condition named after the French physician who identified its main characteristics. One symptom of this condition is an abnormally small lower jaw (micrognathia) with receding chin. Another feature is a displaced tongue, usually falling backwards into the throat (glossoptosis). The tongue often appears large relative to the jaw. In 80% of the cases of this condition, the children also have a U-shaped opening in the roof of their mouths (cleft soft palate). Others may have a high arched palate.
How many children have Pierre Robin Syndrome?

Pierre Robin syndrome is uncommon, affecting approximately one out of every 8,000-30,000 live births.

How do you know if your child has Pierre Robin Syndrome?

There are no prenatal tests for this condition. If a cleft palate is present, it may be identified through an ultrasound during pregnancy. Additionally, there are no special tests to confirm the condition in a newborn child. It is usually determined at birth by clinical examination. In addition to the features noted above, symptoms of Pierre Robin Syndrome may include choking on the tongue and natal teeth.

What causes Pierre Robin Syndrome?

The exact cause of Pierre Robin Syndrome is not known. It has been hypothesized that the initiating defect of this condition is the arrested development of the lower jaw, which is believed to occur prior to the ninth week of fetal development. As a result, the tongue takes a posterior location that impairs the development of the palate. Researchers have also looked at the possibility of maternal viruses and folic acid deficiencies as the cause of this condition but have found no conclusive evidence for these hypotheses. There is no known prevention for Pierre Robin Syndrome and genetic counseling is recommended.

The features of Pierre Robin Syndrome can also be features of a multiple defect syndromes such as Trisomy 18 Syndrome (extra 18th chromosome) or Stickler Syndrome (a disorder of the connective tissue). A child may be tested for these syndromes to see if any other disorder is involved.

How can you help a child with Pierre Robin Syndrome?

Feeding and respiratory difficulties are common. Careful feeding in an upright position using a modified nipple can help prevent choking. Outside of feeding times, infants should be placed in a facedown position. This allows gravity to pull the child’s tongue forward and maintain an open airway. Your child’s respiration should be monitored at all times. Intubation (insertion of a tube to keep the trachea open), tongue-lip adhesion, use of a nasal prong, or tracheostomy (surgical opening into the trachea through the neck) may be necessary to maintain an adequate airway for your child. Feeding and respiratory problems decrease over time as the jaw develops and grows to a more normal size. The jaw may catch-up in growth during your child’s first 2-4 years. Most children develop a normal profile by age 6. Surgery is rarely required on the lower jaw.

A series of surgeries is usually necessary to repair a cleft palate. Therefore, your child’s medical team will usually include his pediatrician and a craniofacial or cleft palate specialist. The surgeries usually begin when your child is 6-19 months of age. The timing depends on many factors including the severity of your child’s condition and the preference of his surgeon. Children with cleft palates are prone to fluid buildup behind the eardrum that can affect their hearing or lead to ear infections. Consequently, ongoing monitoring is recommended. Children with cleft palates may also need speech therapy.
What’s in the future for a child with Pierre Robin Syndrome?

Your child's lower jaw will usually develop slowly over time and can fully correct by adult life. In some cases, however, mental retardation and central nervous system damage can occur from complications resulting from respiratory difficulties. If respiratory and feeding difficulties are adequately managed, your child's prognosis is quite good. Additionally, surgery can correct your child’s cleft palate.

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