

Information about Pierre Robin Sequence/Complex

What is Pierre Robin Sequence/Complex?

Pierre Robin Sequence or Complex (pronounced “Roban”) is the name given to a birth condition that involves the lower jaw being either small in size (micrognathia) or set back from the upper jaw (retrognathia). As a result, the tongue tends to be displaced back towards the throat, where it can fall back and obstruct the airway (glossoptosis). Most infants, but not all, will also have a cleft palate, but none will have a cleft lip.

Over the years, there have been several names given to the condition, including Pierre Robin Syndrome, Pierre Robin Triad, and Robin Anomalad. Based on the varying features and causes of the condition, either “Robin Sequence” or “Robin Complex” may be an appropriate description for a specific patient. Pierre Robin was a French physician who first reported the combination of small lower jaw, cleft palate, and tongue displacement in 1923.

What causes this condition?

The basic cause appears to be the failure of the lower jaw to develop normally before birth. At about 7-10 weeks into a pregnancy, the lower jaw grows rapidly, allowing the tongue to descend from between the two halves of the palate. If, for some reason, the lower jaw does not grow properly, the tongue can prevent the palate from closing, resulting in a cleft palate. The small or displaced lower jaw also causes the tongue to be positioned at the back of the mouth, possibly causing breathing difficulty at birth. This “sequencing” of events is the reason why the condition has been classified as a deformation sequence. For some patients, these physical

characteristics may result from another syndrome or chromosomal condition, however.

How common is this condition?

Robin Sequence/Complex is rather uncommon. Frequency estimates range from 1 in 2,000 to 30,000 births, based on how strictly the condition is defined. In contrast, cleft lip and/or palate occurs once in every 700 live births.

Will future children be affected?

It is important to understand that Robin Sequence/Complex can occur by itself (described as “isolated”) or as a feature of another syndrome. Parents who have had one child with isolated Robin Sequence probably have between 1 and 5% chance of having another child with this condition. There have not yet been enough large-scale studies to make more accurate predictions.

When Robin Sequence/Complex is observed in patients with Stickler Syndrome, Velocardiofacial Syndrome, or Treacher Collins Syndrome, genetic/chromosomal factors will influence whether more affected children will be born. Robin Sequence/Complex also occurs in children with environmentally-induced (“teratogenic”) syndromes such as Fetal Alcohol Syndrome and Fetal Hydantoin Syndrome. It is **extremely important** that an infant born with Robin Sequence/Complex be evaluated by a geneticist, who will thoroughly investigate the possibility of an associated syndrome.

What problems can be expected with this condition?

Robin Sequence/Complex, like most birth defects, varies in severity from child to child. Some

1999

children may have more problems than others. Problems with breathing and feeding in early infancy are the most common. Parents need to know how to position the infant in order to minimize problems (i.e. not placing the infant on his or her back). For severely affected children, positioning alone may not be sufficient, and the pediatrician may recommend specifically-designed devices to protect the airway and facilitate feeding. Some children who have severe breathing problems may require a surgical procedure to make satisfactory breathing possible.

The pediatrician or ear, nose, and throat specialist will also carefully monitor the baby for ear disease. Virtually all children with cleft palate are prone to build-up of fluid behind the eardrum. The placement of ventilation tubes in the eardrums may be recommended to reduce fluid build-up. Since ear infections can cause temporary hearing loss that can affect speech and language development, the infant's hearing should also be monitored from early infancy by an audiologist.

How is the condition treated?

In many patients, the lower jaw (mandible) grows rapidly during the first year of life. In some children, the jaw may grow so quickly that by the time the child is approximately four to six years of age, the profile looks normal. Children who do not experience this "catch-up" growth may require surgery on their jaws. It is not fully understood why children's jaws grow at varying rates.

The cleft palate, if present, needs to be surgically closed. The timing of the surgery depends on the child's growth and development, but it is generally done at 1 to 2 years of age. Because children with cleft palate are at higher risk for delayed or defective speech development, they should be monitored by a speech pathologist throughout early childhood.

Where can children be treated?

Since children with Robin Sequence/Complex may have a variety of health concerns, parents are strongly advised to locate a craniofacial center where evaluation and treatment planning can be coordinated by an experienced multidisciplinary staff composed of health care professionals from many different specialties.

For further information on cleft lip and palate, or for a referral to a cleft palate/craniofacial team, please contact:

**Cleft Palate Foundation
1504 East Franklin Street, Suite 102
Chapel Hill, NC 27514**

**800.24.CLEFT
919.933.9044
919.933.9604 fax
info@cleftline.org
www.cleftline.org**

BIBLIOGRAPHY

- Cohen, MM Jr. (1999). Robin Sequence and Complexes. American Journal of Medical Genetics 84: 311-315.
- Gorlin, RJ, Cohen, MM, Levin, LS (1990) Syndromes of the Head and Neck 3rd Edition. NY: Oxford Univ. Press, pp. 700-704.
- Jones, KL (1997). Robin Sequence. Smith's Recognizable Patterns of Human Malformation 5th Edition. Philadelphia: WB Saunders, p.234.
- Lehman JA, Fishman, JR, Neiman, GS (1995). Treatment of Cleft Palate Associated with Robin Sequence: Appraisal of Risk Factors. Cleft Palate-Craniofacial Journal 32: 25-29.
- Pashayan, HM and Lewis, MB (1984). Clinical Experience with Robin Sequence. Cleft Palate Journal 21: 270-276.
- Sheffield, LJ, Reiss, JA, Gilding, M (1987). A Genetic Follow-Up Study of 64 Patients with Pierre Robin Complex. American Journal of Medical Genetics 28: 25-36.
- A series of invited articles also appeared in: Cleft Palate-Craniofacial Journal, May 1992, Volume 29:3, pp.201-253.