

PIERRE ROBIN SEQUENCE (PRS)

Description

Pierre Robin is a sequence instead of a syndrome because it is caused by a series of *events* that take place in fetal development. Beginning with a defect of the lower fetal jaw (abnormally small jaw or *micrognathia*), the tongue becomes abnormally placed in the back of the throat (*glossoptosis*) and, in turn, causes a cleft palate (hole in the roof of the mouth). The too small lower jaw does not leave room to accommodate the tongue. The tongue therefore accommodates by forming farther back and higher up in the mouth. Not only does this placement of the tongue inhibit the airways, it also prevents the full closure of the developing soft palate; hence the cleft.

Infants with breathing difficulties as a result of tongue placement, also have difficulty swallowing (dysphagia). With swallowing problematic, these children often fail to thrive and may even temporarily stop breathing. In turn, these breathing problems could lead to enlargement of part of the heart, lung malfunctioning, high blood pressure and even congestive heart failure.

Prevalence/Causes

Pierre Robin sequence occurs in 1:8,000 live births with the cause being either genetic or external. Genetically, it can be passed on from a parent either alone or as part of another syndrome. Non-genetic, external causes may be uterine crowding of the fetus that impedes jaw growth or the presence of large fibroids.

Treatment

The main treatment focus is on ensuring that the child's airway remains open when the tendency is for the tongue to block it. Obstructed breathing results in respiratory distress as the baby's lungs and other organs strain to receive enough oxygen. Depending on the individual, surgical intervention may or may not be warranted.

By and large, breathing difficulties tend to be less severe and jaw growth often accelerates as the child becomes older. Laying the baby on its tummy prevents the tongue from falling into the back of the throat in these cases.

Moderate cases of PRS can, at times, be treated by a temporary surgical procedure that attaches the tip of the tongue to the lower lip to prevent it from falling backwards. This procedure is reversed when the lower jaw has grown and is able to keep the tongue in the proper position.

In those cases where breathing is severely blocked, a tracheostomy may be required. This procedure involves creating an opening through the front of the neck into the windpipe (trachea).

Where a cleft palate is present, surgical repair is necessary (see Cleft Lip and Palate). Surgery may also be considered to improve appearance.

Associated Conditions

- Cleft palates can affect the ability to eat and speak properly so that a speech pathologist is often an essential part of the care team.
- Ear infections are also common with clefts.
- Breathing problems can lead to lung malformation and enlargement of part of the heart, high blood pressure and possibly congestive heart failure.
- 40% of infants with Pierre Robin have Stickler syndrome (see additional information)
- 15% of infants have Velo-cardio-facial syndrome (see additional information)