Patient information

Cleft lip and palate

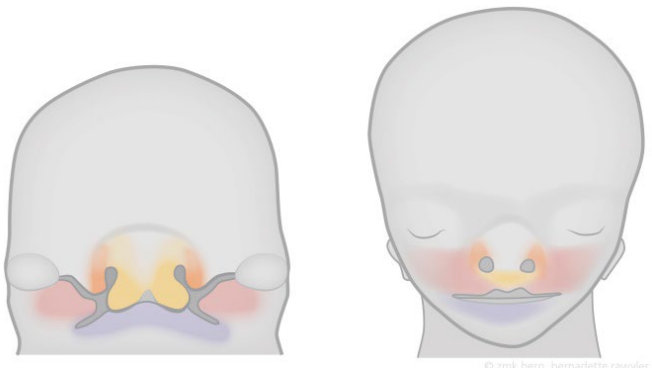
Cleft deformities manifest in a variety of ways. The lips, jaw, and palate may all be affected, either individually or in combination. Cleft palate may additionally involve the hard and/or soft palate. The many different combinations give rise to a variety of individual manifestations. Cleft deformities may generally be divided into three main groups:

- Cleft lip or cleft lip and jaw
  a. complete or incomplete
  b. unilateral or bilateral
- Cleft soft palate or cleft hard and soft palate
  a. complete or incomplete
  b. open or submucous
- Complete cleft lip, jaw and palate
  a. unilateral or bilateral
- Associated malformations

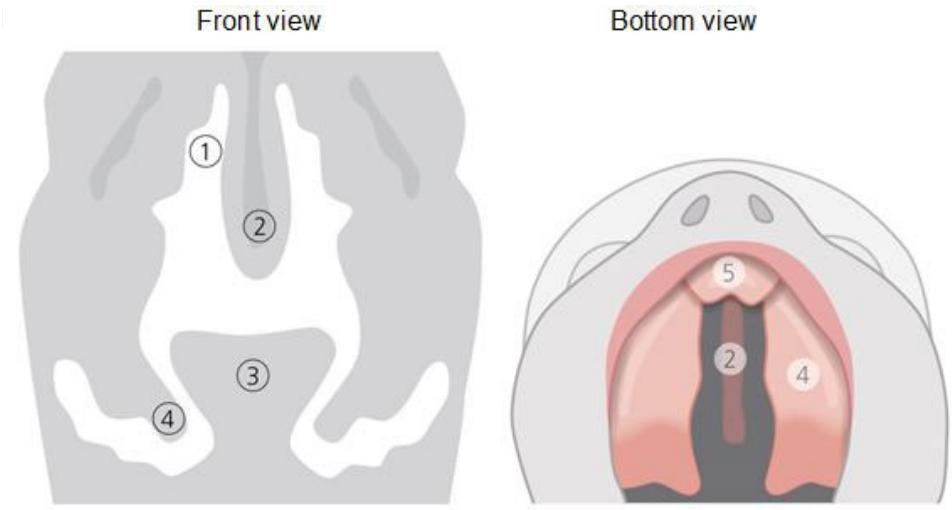
Manifestations

Definition

Approximately one in five hundred newborn babies is affected by cleft deformities of the lip, jaw or palate, although the severity of the manifestations varies. Cleft lip, jaw and palate is therefore one of the most common congenital disorders in humans. Boys are affected more often than girls (3:2). The cause is believed to be a combination of several genetic factors and possibly external, unknown “environmental factors”. Thus, cleft deformities occur more frequently over several generations in some families, although no strict hereditary pattern is identifiable.
The type of the cleft deformity is determined by the point in time at which development is impaired. Impaired development in the 6th to 8th week leads to the formation of cleft lip or cleft lip and jaw. Cleft palate, in contrast, develops from the 9th to 12th weeks.
In general, cleft deformities may be detected from the 20th to 22th weeks of pregnancy by ultrasound, although cleft lip and cleft lip and jaw may be detected with more certainty than isolated cleft palate.

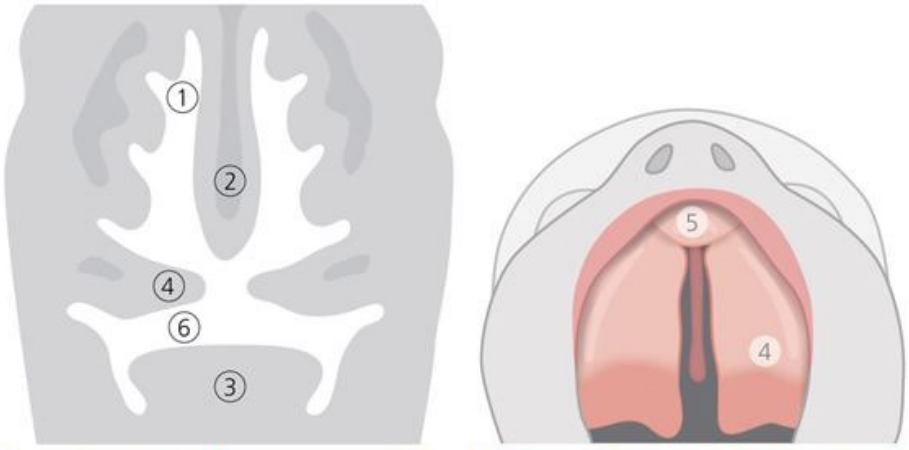


Fusion of the facial prominence: Embryo at 7 weeks and embryo at 10 weeks. Formation of the nostrils by fusion of the medial (yellow) and lateral (orange) nasal prominence at 7 weeks. Formation of the upper lip at 10 weeks by fusion of the medial nostril (yellow) with the right and left upper jaw prominence (red).



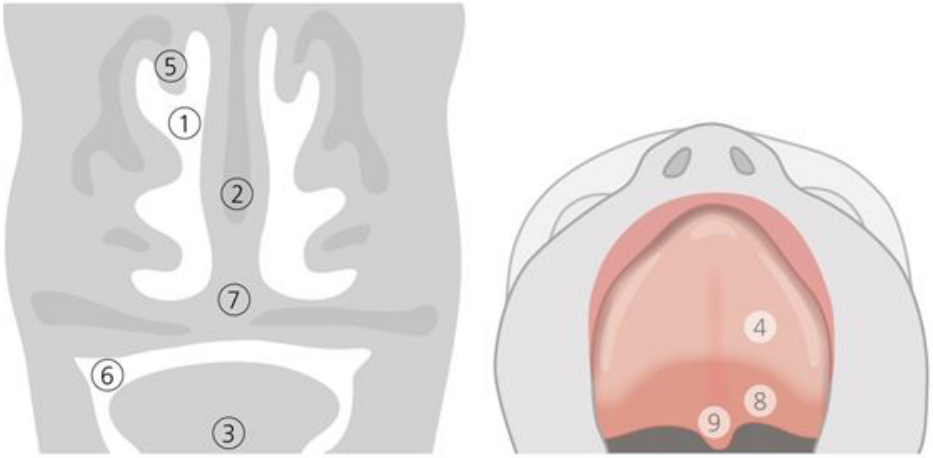
1 Nasal cavity
2 Nasal septum
3 Tongue
4 Palatal process
5 Premaxilla

6½-week embryo. The tongue lies between the palatal processes.



1 Nasal cavity
2 Nasal septum
3 Tongue
4 Palatal process
5 Premaxilla
6 Oral cavity

7½-week embryo. The tongue is displaced downwards, the palatal processes begin moving horizontally.



1 Nasal cavity
2 Nasal septum
3 Tongue
4 Palatal process
5 Premaxilla
6 Oral cavity
7 Fusion of the palatal processes
8 Soft palate
9 Uvula

10-week embryo. The palatal processes are fused to one another and with the nasal septum.

Manifestations

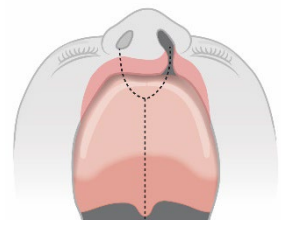
The different types of cleft lip, jaw and palate may be roughly classified into three groups depending on affected regions. A cleft may thereby split the affected region either completely or incompletely. The lip or palate is only partially fused in incomplete separation. The mucosa may remain intact in a cleft palate such that the deformity may not be visible on the outside, but the underlying bones and muscles remain separate. The lip, jaw and hard palate may be affected either on the right or left, or on both sides. The left and right sides may also be affected to variable extents. Thus, clefts in different regions may be variable in severity and may develop on one or both sides.

The cleft appears along the lines of normal tissue growth during development. All humans therefore have a cleft lip and palate at an early point in their development. Should fusion fail to occur within the intended time span, tissue can no longer grow together thereafter and the two halves of the lip or palate remain covered by skin and mucosa. No open wounds are thus seen at birth and everything is covered by skin and mucosa. Despite the cleft, the child has no pain or discomfort after birth. Lip fusion occurs approximately in the 6th to 8th weeks of pregnancy and the palate fuses around the 9th to 12th weeks. Apart from that, though, development before and after is normal. Therefore, in a cleft lip, jaw and palate, uncontrolled development does not occur in the wrong direction, but remains briefly suspended at a decisive point in time. The reason why this occurs in about one in every 500–700 newborn babies usually remains unexplained. Occasionally another family member is also affected, pointing to an interaction of several genes. On the other hand, a single genetic defect is found to be the definitive cause only in very rare cases.

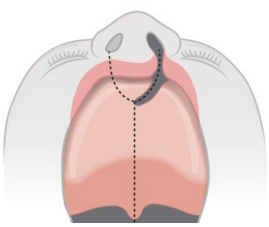
1) Cleft lip or cleft lip and jaw
    a. complete or incomplete
    b. unilateral or bilateral

A cleft deformity in cases of cleft lip is limited to the upper lip and nostril. The cleft appears as an indentation in the red part of the lip or split of the lip, which runs to the opening of the nostril. It may occur on one or both sides. Depending on the extent, the wing of the nose may be laterally displaced. The lateral primary incisor directly adjacent to the cleft may be missing or be present on either side of the cleft.

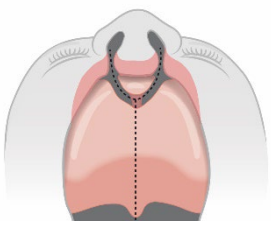
In cleft lip and jaw, the cleft continues into the mouth up to the part of the bony maxilla that holds the teeth (alveolar process). The cleft in this anterior section of the maxilla is called cleft jaw. Among all forms of cleft lip, jaw and palate, this cleft form occurs in about one fifth of cases.



Unilateral cleft lip



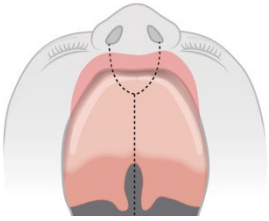
Unilateral cleft lip and jaw



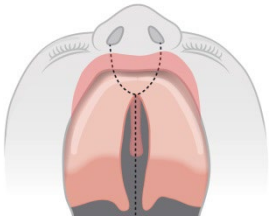
Bilateral cleft lip and jaw (very rare)

2) Cleft soft palate or cleft hard and soft palate
    a. complete or incomplete
    b. open or submucous

Cleft palate may affect either the soft palate, or both hard and soft palates. The hard palate is the immobile anterior part of the palate, which is formed by bone, making it “hard”, and covered by light pink mucosa. The soft palate is the mobile posterior part of the palate, which ends at the uvula. This part is also known as the velum. The interior of the soft palate is composed of muscles. A complete cleft splits the hard or soft palate along its entire length, while an incomplete cleft palate splits only the posterior aspect of the hard or soft palate. Oral mucosa may also remain intact and bone may split only on the inside of the hard palate or muscle may be split only on the inside of the velum. This condition is known as a submucous cleft palate. Cleft palates make up about one third of all clefts.



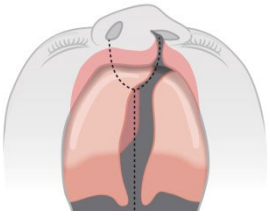
Cleft soft palate



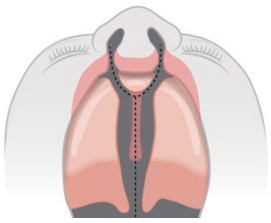
Cleft hard and soft palate

3) Complete cleft lip, jaw and palate
    a. unilateral or bilateral

The most common manifestation is a complete unilateral cleft lip, jaw and palate. Complete unilateral and bilateral clefts comprise about 50% of all cleft deformities and thus occur in approximately one out of every 1000 births. In patients with a complete cleft of the hard palate, the bony nasal floor on the side of the cleft is deficient and the oral and nasal cavities remain connected to each other. The nasal septum is displaced to the healthy side, which distorts midface symmetry. The wing of the nose is flattened to a variable extent depending on cleft width and is displaced inferiorly and laterally. The part of the bony maxilla that holds the teeth (alveolar process) is also affected. In a total bilateral cleft lip, jaw and palate the bony nasal floor is bilaterally deficient and the mid-section of the maxilla that holds the incisors (premaxilla) is positioned far anteriorly. As a result of the forward displacement of the premaxilla, the tissue bridge in between the nostrils (columella) is shortened and the tip of the nose appears flattened.



Complete unilateral cleft lip, jaw and palate



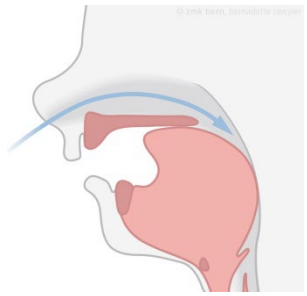
Complete bilateral cleft lip, jaw and palate

Associated malformations
Malformations associated with cleft lip, jaw and palate are not unusual. With an isolated cleft palate, additional malformations occur in about 20% of affected children. This is termed a malformation syndrome if the clinical presentation of these malformations is typical. We work together with the clinical genetics team and the pediatric clinic to provide a more thorough evaluation and counseling in this field.

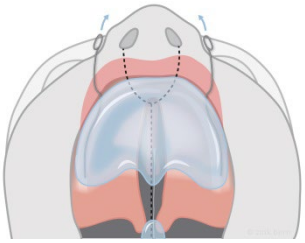
Among combined malformations, the Pierre Robin sequence occurs most frequently and affects about one in every 8000 births. The Pierre Robin sequence is characterized by a very small mandible, which may be identified by a receding chin, a posteriorly displaced tongue and a cleft palate. It is assumed that the undersized mandible pushes the tongue upwards which does not allow the palate to close, leading to a sequence of malformations.

The tongue may fall backwards in the supine position due to the receding mandible. The airway maybe occluded because the tongue obstructs the nasopharynx and the oropharynx. This may result in an oxygen deficiency, which may have severe consequences for the infant. Children frequently choke due to the posterior displaced tongue and often have difficulty coordinating sucking, swallowing and breathing. These children are admitted to neonatal intensive care units to secure the airways and for observation. In intensive care, one of the parents may stay with the child at all times.

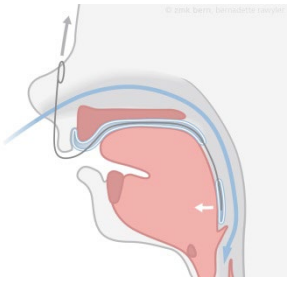
A special palatal plate is usually made for these children in their first days of life. This plate is named after Margrit Bacher from Tübingen, who developed this method. This Tübinger plate (also called Bacher plate) prevents the tongue from falling backwards, enabling the child to breath better through the mouth and nose. As a result, the child does not need to be placed constantly on the stomach to be able to breathe freely. The movement of the tongue into its correct position is stimulated, enabling development of correct movement patterns.



Pierre Robin sequence with restricted airway due to the posteriorly displaced tongue.



Insertion of the palatal plate extending to the base of the tongue and with outward extensions.



By pulling on the outer extensions, the base of the tongue is pressed forward and the airway is opened.

Nevertheless, swallowing may initially be incorrect due to the small mandible and the posterior position of the tongue, with a risk that the child may choke on food. The risk of choking is also increased because children often suffer from reflux. Nutrition is developed with the support of speech therapy for these reasons.
Breathing and feeding problems usually normalize completely on their own in the first 3–6 months of life, so the special palatal plate is no longer needed. The timing of surgery for cleft palate depends on the child's weight, width of the palatal cleft and the extent of recession of the mandible, and is usually performed in infants aged 8 months.
Respiratory problems rarely do not fully improve despite treatment with the plate and palatal closure. The mandible must be surgically extended (distraction) in such cases.