Lec. 12 Cleft lip and palate
Part 1

Clefts involving the lip and/or palate (CLP) or isolated clefts of the palate (CP) are the most common congenital anomaly to affect the craniofacial region in man, comprising about 65% of all anomalies affecting the head and neck. They represent a complex phenotype and reflect a failure of the normal mechanisms involved during early embryological development of the face. In human populations, CLP and CP can be broadly subdivided into:

• Nonsyndromic, which occur in isolation; and

• Syndromic, which occur in combination with other physical and developmental anomalies.

Clefts of the lip and palate

The prevalence of cleft lip and palate varies geographically and between different racial groups. Among Caucasians, this anomaly occurs in approximately 1 in every 700 live births and the prevalence is increasing. A family history can be found in around 40% of cases of cleft lip with or without cleft palate, and the risk of unaffected parents having another child with this anomaly is 1 in 25 (Box 1). Males are affected more frequently than females, and the left side is involved more commonly than the right. Interestingly, the severity of the cleft is usually more marked when it arises in the less common variant.

Box 1: Genetic risks of cleft lip and palate

• Parents with no cleft but with one affected child: risk for next child = 1 in 25 (4%).

• One parent with cleft lip and palate: risk for first child = 1 in 50 (2%).

• One parent with cleft lip and palate and first child with cleft lip and palate: risk for next child = 1 in 10 (10%).

• Both parents affected: risk for first child = 3 in 5 (60%).

Isolated cleft of the secondary palate

Isolated cleft occurs in around 1 in 2000 live births and affects females more often than males. Clefts of the secondary palate have a lesser genetic component, with a family history in around 20% and a reduced risk of further affected offspring to normal parents (1 in 80). Isolated cleft palate is also found as a feature in a number of syndromes including Down, Treacher–Collins, Pierre–Robin, and Klippel–Fiel syndromes.

Aetiology

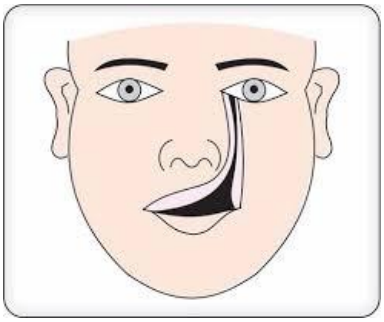
In normal development, fusion of the embryological processes that comprise the upper lip occurs around the sixth week of intrauterine life. ‘Flip-up’ of the palatal shelves from a vertical to a horizontal position followed by fusion to form the secondary palate occurs around the eighth week. Before fusion can take place, the embryological processes must grow until they come into contact. Then breakdown of the overlying epithelium is followed by invasion of mesenchyme. If this process is to take place successfully, a number of different factors need to interact at the right time. Evidence from population studies and experimental data suggests that both genetic and environmental factors play a part in the aetiology of clefts. Specific gene mutations have been shown to be linked to cleft lip and/or cleft palate. Environmental factors that have been implicated include anticonvulsant drugs, folic acid deficiency, and maternal smoking.

It is postulated that isolated cleft palate is more common in females than males because transposition of the palatal shelves occurs later in the female fetus. Thus, greater opportunity exists for an environmental insult to affect successful elevation, which is further hampered by widening of the face as a result of growth in the intervening period. At the embryological level, perturbations in a variety of mechanisms during facial development are known to cause clefting (Figure 1).

A number of possible causes of cleft lip and palate have been identified, including exposure to some teratogens. Insults to developing tissues usually precede the steps that culminate with closure of the lip and palate, and it is interesting that maternal smoking is a definite risk factor.

In addition to “typical” cleft lip and cleft palate, unusual facial clefts occur that also result from the failure of facial prominences to properly form or unite. Examples include macrostomia, a defect at the junction of the maxillary and mandibular prominences that may result from a growth deficiency in either or both of these growth centers, and oblique facial clefts that occur at the junction of the maxillary growth center with either the lateral nasal or maxillary growth center.

macrostomia



oblique facial cleft

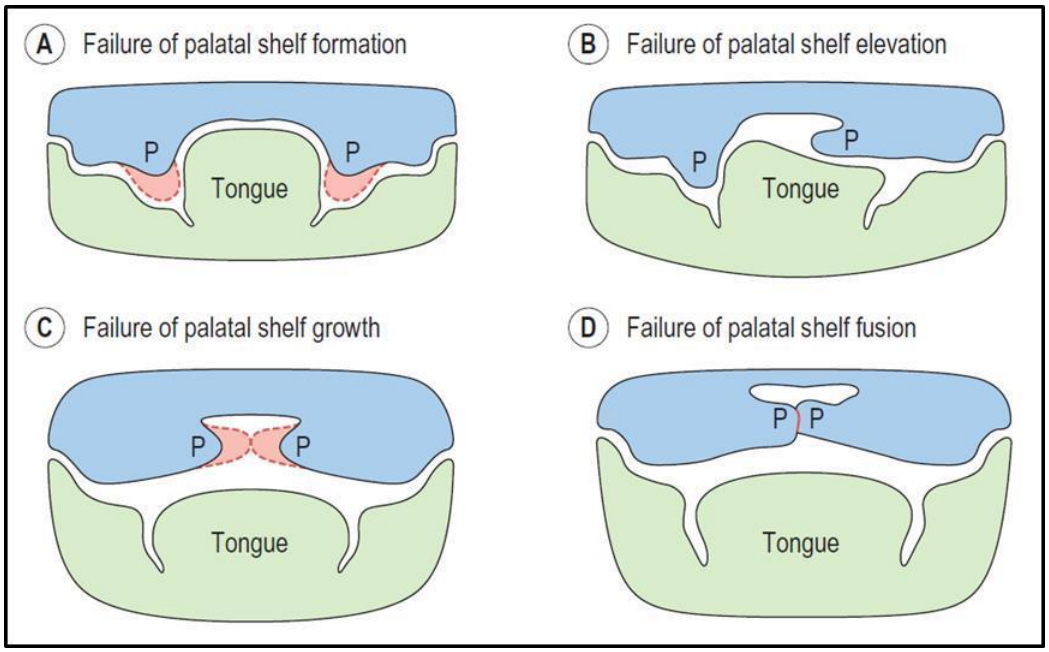


Figure 1: Embryonic causes of cleft palate.

Classification

A number of formal classifications have been described for CLP and CP; however, the clinical presentation of these conditions is so variable that a specific description of each individual case is more useful (Figure 2).

• CLP can range from simple notching or isolated clefting of the upper lip with or without involvement of the alveolus, to complete unilateral or bilateral clefts of the lip, alveolus and hard/soft palate; and

• CP can range from a simple submucous cleft (a lack of continuity of the muscles across the palate) or bifid uvula to a complete cleft involving the primary and secondary palate.

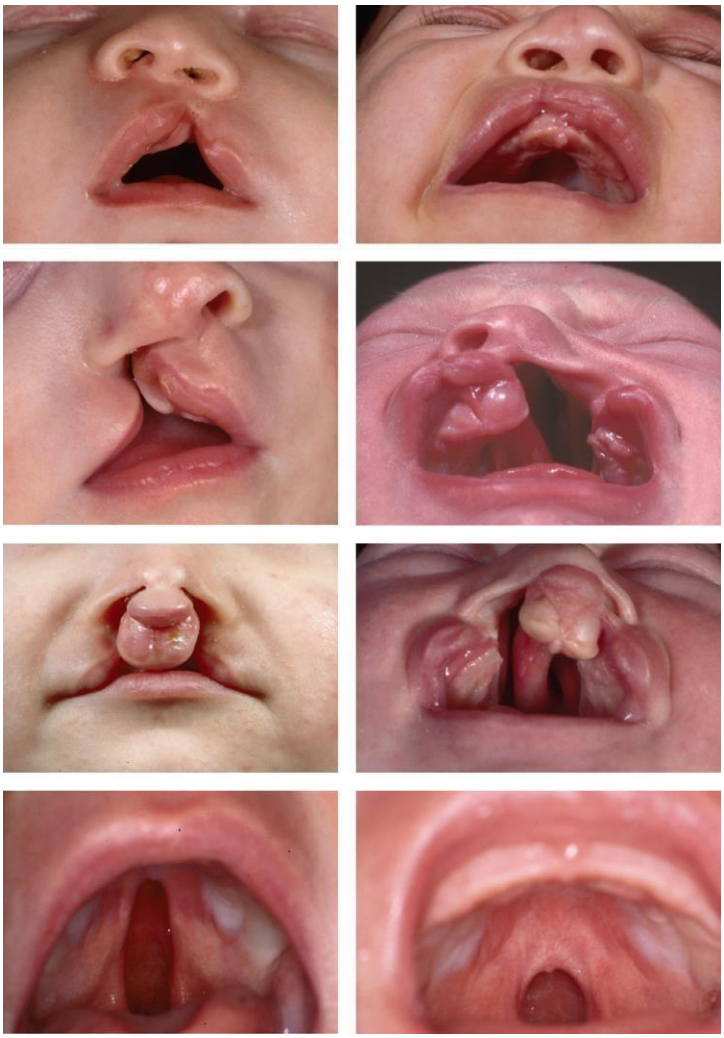


Figure 2: Orofacial clefting. Unilateral cleft lip (top row), unilateral cleft lip and palate (second row), bilateral cleft lip and palate (third row), isolated cleft palate (bottom row).

Treatment

A child born with orofacial clefting will require complex long-term treatment, depending upon the severity of the cleft, and there may be life-long implications for some of those patients. The principal objectives of treatment are to establish:

• Good facial appearance;
• Good orofacial function during speech, eating and swallowing;
• An aesthetic, functional and stable occlusion; and
• Good hearing.

If these objectives are achieved, they maximize the chances of an affected child growing up and developing normally within their social environment. The clinical management of children born with clefting is most effective when carried out by a fully integrated team, in a centralized unit that treats a high number of patients. The modern cleft team therefore includes a number of key members, in addition to other specialists who may be involved with long-term care including: Cleft surgeon, Orthodontist, Speech therapist, Cleft nurse, Ear–nose–throat surgeon, Paediatrician, Paediatric dentist, Restorative dentist, Psychologist, Paedodontist, Audiologist, Geneticist, General dental practitioner and Nutritionist. Orthodontic intervention will usually be required at several time points during the first two decades of an affected child's life, often to facilitate the intervention of other specialities.

Problems in management

Congenital anomalies:

The disturbances in dental and skeletal development caused by the clefting process itself depend upon the site and severity of the cleft.

Lip only:

There is little effect in this type, although notching of the alveolus adjacent to the cleft lip may sometimes be seen.

Lip and alveolus:

A unilateral cleft of the lip and alveolus is not usually associated with segmental displacement. However, in bilateral cases the premaxilla may be rotated forwards. The lateral incisor on the side of the cleft may exhibit some of the following dental anomalies:

• Congenital absence

• An abnormality of tooth size and/or shape

• Defects of the enamel

• Or present as two conical teeth, one on each side of the cleft.

Lip and palate:

In unilateral clefts, rotation and collapse of both segments inwards anteriorly is usually seen, although this is usually more marked on the side of the cleft (the lesser segment). In bilateral clefts, both lateral segments are often collapsed behind a prominent premaxilla (Fig. 3).

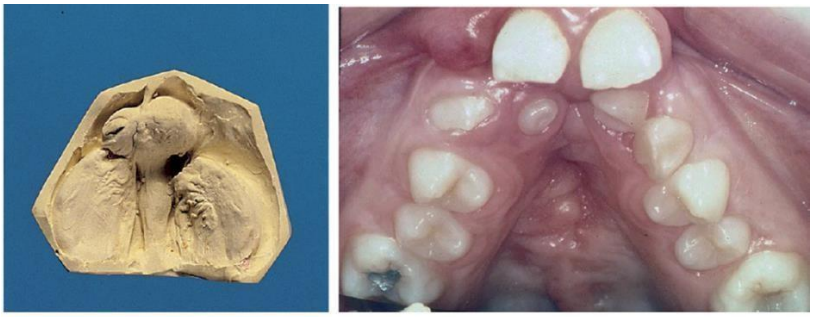


Figure 3: (a) Upper model of a bilateral complete cleft lip and palate showing the inward collapse of the lateral segments behind the pre-maxillary segment; (b) upper arch of a patient in the late mixed dentition with a bilateral complete cleft lip and palate.

Palate only:

A widening of the arch posteriorly is usually seen. It has been shown that individuals with a cleft have a more concave profile, and while a degree of this is due to a restriction of maxillary growth, research indicates that cleft patients have a tendency towards a more retrognathic maxilla and mandible and also a reduced upper face height compared with the normal population.