Cleft Lip and Cleft Palate Features and Management

Cleft lip and cleft palate, collectively known as **orofacial clefts**, are congenital anomalies that occur due to improper fusion of the maxillofacial structures during embryonic development, typically between the **4th and 12th weeks of gestation**.

These anomalies result in a gap or discontinuity in the lip and/or palate and can occur as:

- **Cleft lip (CL)**: may be unilateral or bilateral; complete or incomplete
- **Cleft palate (CP)**: may involve only the soft palate or extend through both hard and soft palates
- **Cleft lip and palate (CLP)**: combination of both
- **Atypical clefts**: median, transverse (lateral), and oblique facial clefts (rare)

Epidemiology

- **Prevalence**: Approximately **1 in every 500–550 live births** is affected globally.
- **Ethnic variation** (per 1000 live births):
  - Asians: **1.61**
  - Caucasians: **0.9**
  - African descent: **0.31**
- **Sex distribution**:
  - **Cleft lip (± cleft palate)** is more common in **males**
  - **Isolated cleft palate** is more common in **females**
- Cleft lip with or without cleft palate (CL ± P) is considered **genetically distinct** from isolated cleft palate (CP).

Etiology and Pathogenesis

Multifactorial causes include:

- **Genetic mutations** (e.g., IRF6, MSX1, PVRL1)
- **Teratogens**: alcohol, anticonvulsants (phenytoin), retinoic acid, tobacco
- **Nutritional deficiencies**: especially **folic acid deficiency**
- **Syndromic associations**: over 400 syndromes (e.g., Van der Woude, Pierre Robin sequence)

Clinical Features

Cleft Lip

- May be **unilateral or bilateral**, **complete** (extends into the nostril) or **incomplete** (limited to the lip).
- Often visible at birth and may involve underlying alveolar ridge.

Cleft Palate

- Can involve **soft palate**, **hard palate**, or both.
- May appear as a **V-shaped** or **horseshoe-shaped** defect.
- **Feeding and speech difficulties** are hallmark signs.

Pierre Robin Sequence

- Triad:
  - **Micrognathia** (small jaw)
  - **Glossoptosis** (tongue falls backward)
  - **Cleft palate**
- Associated with airway obstruction and feeding difficulties.
- Often linked to **congenital heart defects** (prevalence up to **15%** in PRS patients).

Associated Congenital Abnormalities

- **Congenital heart disease**
- **Hearing loss** due to **recurrent otitis media**
- **Dental malocclusion** and enamel hypoplasia
- **Speech and language delays**
- **Facial asymmetry**

Diagnosis

- **Prenatal ultrasound** (as early as 20 weeks) can detect larger clefts.
- **Postnatal assessment** includes a thorough physical exam, feeding evaluation, hearing screening, and genetic workup if syndromic features are present.

Initial Neonatal Management

Key priorities:

- **Ensure adequate nutrition** (prevent aspiration and malnutrition)
- **Manage airway** in cases of glossoptosis
- **Prevent infections**, particularly otitis media
- **Family support and counseling**

Feeding Support

- **Isolated CL**: Breastfeeding often possible with proper latching techniques.
- **Cleft Palate**: Use of **specialized bottles** (Haberman feeder, cross-cut nipple).
- Position infant **upright** during feeding to reduce nasal regurgitation and aspiration.
- **Palatal obturators** may be used to facilitate feeding.

Multidisciplinary Team Approach

Management requires collaboration among:

- Pediatrician
- Plastic surgeon
- Otolaryngologist (ENT)
- Audiologist
- Speech-language pathologist
- Pediatric dentist/orthodontist
- Geneticist
- Psychologist
- Cleft/craniofacial nurse coordinator

Treatment Timeline and Protocol

Age	Intervention
Birth	Diagnosis, parental counseling, feeding support, genetic evaluation
3 months	Surgical **cleft lip repair** (cheiloplasty), **tympanostomy tubes** if otitis media present
6 months	**Orthodontic assessment**, baseline **speech evaluation**
9-12 months	Surgical **cleft palate repair** (palatoplasty), continued speech therapy
1–7 years	**Orthodontic treatment**, ongoing speech therapy
7–8 years	**Alveolar bone grafting** (to support tooth eruption and nasal floor)
>8 years	**Orthodontics continues**, additional surgeries as needed

Surgical Considerations

- **Rule of 10s**: traditional criteria for lip repair:
  - **10 weeks old**,
  - **10 pounds (4.5 kg)**,
  - **Hemoglobin ? 10 g/dL**
- Lip repair (cheiloplasty) improves feeding, speech development, and psychosocial outcomes.
- Palate repair (palatoplasty) enhances velopharyngeal function critical for speech.
- Bone grafting restores alveolar continuity to support teeth.

Complications and Long-Term Outcomes

- **Hearing loss** due to eustachian tube dysfunction
- **Speech delays** (hypernasal speech, articulation errors)
- **Psychosocial effects**: body image issues, bullying

- **Dental malocclusion** and nasal deformities requiring revision surgery

Prognosis

With early intervention and multidisciplinary care, most children with orofacial clefts have **normal development, speech, and psychosocial outcomes**.

High-Yield

- Cleft lip/palate can be diagnosed **prenatally** with **ultrasound**.
- Feeding difficulty is more severe in **cleft palate** than in **cleft lip**.
- **Pierre Robin sequence** is associated with airway obstruction and **cardiac defects**.
- Use **special bottles** and position upright to prevent **aspiration**.
- **Surgery begins around 3 months** for cleft lip and **9–12 months** for cleft palate.
- Always consider **associated anomalies**; refer for **audiologic** and **speech evaluations**.
- **Genetic counseling** is important due to familial recurrence risks.