

Cleft Lip / Palate



This guideline was updated in August 2011 by Dr Saul Snowwise, with input from members of the New Zealand Maternal Fetal Medicine Network.



Background

Facial clefts can be a difficult diagnosis to make and difficult for affected families to deal with.

Accurately diagnosing facial clefts and discovering other potential associated abnormalities are important as both factors affect prognosis.

Objective

To provide guidance and a consistent approach for the accurate diagnosis and management of mothers and fetuses presenting with cleft lip and/or palate.

Definition

Facial clefts are deformations of the face caused by relative tissue deficits along linear anatomical planes.

Differential Diagnosis

- Normal Philtrum
- Amniotic bands
- Facial mass
 - Teratoma
 - Frontal encephalocele
 - Hemangioma
- Holoprosencephaly
- Trisomy 13/18
- Syndrome (over 300 with facial cleft as part)



Incidence

Cleft Lip: 0.15 % of live births

80% of cleft lip has an associated cleft palate

- Ethnic variation
 - 1:600 Asians
 - 1:1,100 Caucasians
 - 1:2,500 African Americans
- Gender variation
 - M > F
- 30% syndromic

Isolated Cleft Palate:

- Constant prevalence across races
- F > M
- 50% syndromic

Classification

Type I Unilateral cleft lip

Type II Unilateral cleft lip and palate

Type III Bilateral cleft lip and palate

Type IV Midline cleft lip and palate

**Type I – III result from failure of lip/palate fusion

**Type IV results from agenesis of the intermaxillary process



Aetiology

Multifactorial with both genetic and environmental factors

Genetic defects – Sonic Hedgehog, TGF-alpha

- variant, TGF-beta-3, IRF-6 just some of genes implicated in CL/P
- Medications – retinoic acid, valproic acid, hydantoin, Methotrexate
- Cigarette smoking
- Alcohol
- Folate deficiency
- Maternal obesity

Diagnosis

- Not readily seen until about 13-14 weeks gestation when soft tissue on the face has developed sufficiently.
- “snout view” – angled, coronal, nose-mouth view is best for identifying on 2D scan
- 3D scan helpful with surface and volume rendering

Detection Rate

Isolated Cleft Lip	20% detected antenatally
Cleft Lip and Palate	33% detected antenatally
Isolated Cleft Palate	0.3% detected antenatally

higher detection rates when other anomalies identified

3D scan and MRI both increase detection



Associated Abnormalities

Cleft location important for predicting associated abnormalities

Unilateral cleft lip/palate	10% associated anomalies
Bilateral cleft lip/palate	25% associated anomalies
Midline cleft lip/palate	100% associated anomalies

Prognosis

Excellent overall prognosis for isolated cleft lip and palate

Trisomies, syndromic infants, and infants with associated anomalies have worse prognosis dependent on presence and extent of specific abnormalities.

On-going Management

1. Detailed anatomy scan to exclude associated abnormalities
2. Amniocentesis should be offered to all patients with plan of care according to results
 - a. Unilateral CL/P has 20% Aneuploidy risk
 - b. Bilateral CL/P has 30% Aneuploidy risk
 - c. Midline CL/P has 50% Aneuploidy risk
3. Preparation for delivery
 - a. Plastic surgeon, ENT, Dentist, speech and language therapist, social work, psychiatrist, genetic counsellor
4. Surgery
 - a. Repair of cleft lip at 2-3 months
 - b. Repair of cleft palate at 9-18 months



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