CLEFT LIP AND/OR PALATE

Otherwise known as?

Cleft lip and/or palate is the usual umbrella term, but may also be described as an orofacial cleft. Depending on the structures affected, it may be described more specifically as:

- Cleft lip
- Cleft palate
- Cleft lip and palate

Signs & Symptoms

Cleft lip and/or palate occurs when parts of the upper lip, gum and/or roof of the mouth do not join completely during early development. This leaves a gap or opening that is present at birth.

Signs and symptoms vary depending on the severity of the condition.

1. Signs of Cleft Lip

- A visible split or gap in the upper lip, which may be:
  - Unilateral (one side) or bilateral (both sides)
  - Small (a notch) or large (extending to the nose)
- Flattened or asymmetrical nose shape (if the cleft affects the nostril area).
- Difficulty with sucking and feeding, especially in newborns.

2. Signs of Cleft Palate

- An opening in the roof of the mouth (hard or soft palate).
- Difficulty feeding, as the baby may struggle to create suction.
- Nasal-sounding speech due to air escaping through the nose.
- Frequent ear infections and possible hearing loss.
- Dental problems, such as missing, extra, or misaligned teeth.

(continued overleaf)

Craniofacial Australia
204 Melbourne Street
North Adelaide
South Australia, 5006

W: craniofacial.com.au
E: familysupport@acmff.org.au
P: (08) 8267 4128

Causes (*the why*)

Cleft lip and/or palate occur when the tissues that form the lip and palate fail to fuse properly during early foetal development (usually between 4-10 weeks of pregnancy). The exact cause is often unknown, but several genetic and environmental factors can contribute.

1. Genetic Causes

- Family History (Hereditary Factors): If a parent or close relative has had a cleft, the baby may have a higher risk.
- Syndromes & Genetic Conditions: Clefts can occur as part of genetic syndromes, such as:
  - Van der Woude Syndrome
  - Pierre Robin Sequence
  - Treacher Collins Syndrome

2. Multifactorial Causes

- In many cases, cleft lip and/or palate result from a combination of genetic and environmental factors rather than a single cause.

While clefts cannot always be prevented, prenatal care, adequate nutrition (especially folic acid intake), and avoiding harmful substances during pregnancy may help reduce the risk.

Testing & Diagnosis

Cleft lip and/or palate can be diagnosed before birth (prenatally) or after birth (postnatally) through various tests and examinations.

1. Prenatal Diagnosis (Before Birth): doctors may detect cleft lip and/or palate during pregnancy using:

a. Ultrasound (18-22 Weeks Gestation)

- A detailed foetal ultrasound can often identify a cleft lip.
- Cleft palate alone may be harder to detect on ultrasound.

(continued overleaf)

Craniofacial Australia

204 Melbourne Street
North Adelaide
South Australia, 5006

W: craniofacial.com.au
E: familysupport@acmff.org.au
P: (08) 8267 4128

b. Amniocentesis & Genetic Testing (Optional)

- If a cleft is detected on ultrasound, genetic testing may be done to check for underlying syndromes (e.g., Van der Woude syndrome).
- Amniocentesis (sampling amniotic fluid) helps detect chromosomal differences that might be linked to cleft conditions.

2. Postnatal Diagnosis (After Birth): if a cleft lip and/or palate is not detected before birth, doctors will diagnose it at birth through:

a. Physical Examination

- Cleft Lip: A visible gap or split in the upper lip.
- Cleft Palate: An opening in the roof of the mouth, which may be harder to detect if it is a submucous cleft (hidden beneath the skin).

b. Imaging Tests (if needed)

- X-rays or CT Scans: Used in some cases to assess the severity of the cleft, particularly in the palate and skull.

3. Additional Evaluations (for associated conditions)

Since cleft lip and palate can be part of a genetic syndrome, further assessments may be needed:

- Hearing Tests: To check for hearing loss due to ear infections.
- Speech and Feeding Assessments: To identify difficulties with sucking, swallowing, or speaking.
- Dental and Orthodontic Exams: To evaluate tooth development and alignment.

Treatment

Treatment for cleft lip and/or palate involves a multidisciplinary approach, including surgery, speech therapy, dental care, and other supportive treatments. The goal is to improve appearance, speech, feeding, and overall quality of life.

1. Surgical Treatment

Surgery is the primary treatment to repair the cleft and restore function. The timing and type of surgery depend on the severity of the cleft.

(continued overleaf)

Craniofacial Australia

204 Melbourne Street
North Adelaide
South Australia, 5006

W: craniofacial.com.au
E: familysupport@acmff.org.au
P: (08) 8267 4128

a. Cleft Lip Repair (Cheiloplasty)

- Performed at: 3-6 months of age.
- Procedure:
  - The surgeon closes the gap in the lip and reshapes the nose if necessary.
  - May require additional surgeries to address appearance and improve function.

b. Cleft Palate Repair (Palatoplasty)

- Performed at: 9-18 months of age.
- Procedure:
  - The surgeon closes the opening in the roof of the mouth, helping with speech and feeding.
  - This may also help prevent ear infections and hearing loss.
  - Some children may need a second surgery as they grow.

c. Additional Surgeries (if needed)

- Speech Surgery: For some children with speech difficulties.
- Bone Graft Surgery (Alveolar Bone Graft, 7-12 years old): To fix gaps in the upper jaw and support permanent teeth.
- Orthognathic (Jaw) Surgery (Teenage Years): To address jaw misalignment.
- Nasal & Lip Revision Surgery: To refine appearance and function as the child grows.

2. Feeding Support

Babies with cleft lip/palate often struggle with feeding, so they may require:

- Special bottles/nipples.
- Feeding therapy with a specialist if feeding difficulties persist.

3. Speech Therapy

- Many children with cleft palate have speech difficulties due to improper airflow.
- Speech therapy helps improve clarity and pronunciation.
- Some may need additional speech surgery if therapy alone is not effective.

4. Dental and Orthodontic Care

Children with cleft palate often have dental issues such as missing, extra, or misaligned teeth. Treatment includes:

- Early dental care to prevent cavities.
- Braces or orthodontic treatment to align teeth and address bite problems.
- Prosthetic dental appliances if teeth are missing.

(continued overleaf)

Craniofacial Australia

204 Melbourne Street
North Adelaide
South Australia, 5006

W: craniofacial.com.au
E: familysupport@acmff.org.au
P: (08) 8267 4128

5. Hearing and Ear Care

- Children with cleft palate are prone to ear infections and hearing loss due to Eustachian tube dysfunction.
- Treatment includes:
  - Ear tubes (Tympanostomy tubes) to drain fluid and prevent infections.
  - Regular hearing tests to monitor for any hearing loss.

6. Psychological and Emotional Support

- Children with clefts may face self-esteem issues due to appearance differences.
- Counselling and support groups can help with confidence and emotional well-being.

Long-Term Care and Follow-Up

- Most children with cleft lip and/or palate require ongoing medical care into adolescence.
- A cleft team (including surgeons, dentists, speech therapists, and psychologists) works together to ensure the best possible outcome.

Did you know?

In Australia, approximately 19 babies per 10,000 births are diagnosed with an orofacial cleft (i.e., cleft lip and/or palate)*

Some historians believe that Julius Caesar, the Roman general and statesman, may have had a cleft lip due to the distinctive appearance of his upper lip seen in statues and coins. However, this is purely speculative, and there is no medical confirmation.

References:

*Junaid, M., Slack-Smith, L., Wong, K., Bourke, J., Baynam, G., Calache, H., & Leonard, H. (2022). Epidemiology of Rare Craniofacial Anomalies: Retrospective Western Australian Population Data Linkage Study. The Journal of Pediatrics, 241, 162-172.e169 https://doi.org/10.1016/j.jpeds.2021.09.060

Craniofacial Australia

204 Melbourne Street
North Adelaide
South Australia, 5006

W: craniofacial.com.au
E: familysupport@acmff.org.au
P: (08) 8267 4128

How we can support you:

- Care packs
- Financial assistance
- Family support coordinator
- Connection to other families