APERT SYNDROME


Pictured: Apert-Warrior Daisy

If your child has just been diagnosed with Apert syndrome, you may be feeling overwhelmed. You are not alone.

Apert syndrome is also known by several other names, although "Apert syndrome" is the most widely recognised.

Some alternative terms or descriptions include:

1. Craniofacial dysostosis – This refers to differences in growth of the skull bones, which is a key feature of Apert syndrome.
2. Apert's syndrome – A slight variation in spelling but refers to the same condition.
3. Acrocephalosyndactyly type 1 – this is a descriptive term that highlights two major features of Apert syndrome:
   - Acrocephaly (a cone-shaped head due to early closure of the skull sutures).
   - Syndactyly (fusion of fingers and toes).

These names all refer to the same condition, which is characterized by cranial and facial differences, as well as syndactyly (fusion of fingers and/or toes). The most common and widely accepted term remains "Apert syndrome."

Signs & Symptoms

Apert syndrome is a rare genetic condition characterized by distinctive physical features and, for some people, varying degrees of intellectual disability. Some common signs and symptoms are:

1. Craniofacial Differences:

- Craniosynostosis: Premature fusion of skull bones leading to differences in skull and facial development.
- Midface Hypoplasia: Underdeveloped midface, giving a flattened appearance.
- Protruding Eyes: Eyes that appear to bulge out due to shallow eye sockets.

2. Hand and Foot Differences:

- Syndactyly: Fused or webbed fingers and toes.
- Bony Fusion: Bones in the hands and feet may be fused together.

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3. Dental Issues:

- Malocclusion: Misalignment of teeth due to differences in jaw development.
- Crowded Teeth: Teeth may be crowded due to insufficient space in the mouth.
- High arched palate: The roof of the mouth may be higher and narrower than usual, which can contribute to crowding and bite differences.

4. Other Possible Features:

- Intellectual Disability: Varies widely in severity.
- Ear Differences: Small differences in the shape of the ears.
- Skin Problems: Skin may be thickened or ridged, particularly on the palms and soles.

The severity of these symptoms can vary widely among individuals with Apert syndrome. Treatment typically involves a multidisciplinary approach to address the various medical, surgical, and developmental needs of the affected person.

Causes

Apert syndrome is caused by mutations in the FGFR2 (Fibroblast Growth Factor Receptor 2) gene. This gene plays a crucial role in regulating bone development and growth, particularly in the skull, hands, and feet.

Causes of Apert Syndrome:

1. Genetic Mutation:

- Apert syndrome results from a spontaneous (de novo) mutation in the FGFR2 gene, meaning it usually occurs randomly during early foetal development without being inherited from parents.
- In rare cases, it can be inherited in an autosomal dominant pattern, meaning a parent with the mutation has a 50% chance of passing it on to their child.

2. Effect of the Mutation:

- The FGFR2 mutation causes the premature fusion of skull bones (craniosynostosis), leading to differences in skull and facial development.
- It also affects limb development, leading to webbed or fused fingers and toes (syndactyly).

While the exact reason why a mutation occurs spontaneously is often not fully understood, it is known to be a rare genetic event.

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Testing & Diagnosis

1. Physical Examination

- A doctor will examine the child for characteristic features of Apert syndrome including:
  - Different head shape due to craniosynostosis
  - Syndactyly (webbed or fused fingers and toes)
  - Facial features, such as a flat midface and bulging eyes

2. Imaging Tests

To assess the child's bone structure and confirm craniosynostosis, doctors may use:

- X-rays: to check skull and limb differences.
- CT Scan (Computed Tomography): provides detailed images of skull bones and fusion patterns.
- MRI (Magnetic Resonance Imaging): evaluates brain development and any potential complications, such as hydrocephalus (i.e., fluid build-up in the brain).

3. Genetic Testing

- A DNA test (usually from a blood sample) is used to look for changes (mutations) in the FGFR2 gene, which can confirm the diagnosis. Different gene changes are associated with slightly different patterns of features (sometimes described as subtypes).
- This test helps differentiate Apert syndrome from other similar conditions, such as Crouzon syndrome or Pfeiffer syndrome.

4. Prenatal Diagnosis (Optional)

- If there is a family history or suspected case, genetic testing can be done during pregnancy using:
  - Amniocentesis (sampling amniotic fluid)
  - Chorionic Villus Sampling (CVS) (sampling placental tissue)
- Ultrasound may also detect craniosynostosis or limb differences in the foetus.

5. Additional Evaluations

Since Apert syndrome can affect multiple systems, other specialists may conduct:

- Hearing tests (audiology exam)
- Eye exams (ophthalmology evaluation)
- Cognitive and developmental assessments

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Treatment

There is no cure for Apert syndrome, but treatment focuses on managing symptoms and improving quality of life. A multidisciplinary approach involving surgeons, geneticists, paediatricians, and therapists is essential.

1. Surgical Treatments

Surgery is the primary treatment for Apert syndrome to address skull, facial, hand, and foot differences and function.

a. Skull Surgery (to address craniosynostosis)

- Goal: To allow normal brain growth and reduce pressure inside the skull.
- Procedure: Cranial vault remodelling (i.e., surgery to reshape the skull) performed between 6-12 months of age.
- Benefits: Improves skull shape, brain development, and prevents complications like increased intracranial pressure.

b. Facial Surgery

- Midface advancement (Le Fort III osteotomy) is usually performed around 6-10 years old to address midface underdevelopment and improve breathing.
- Jaw surgery may be needed later to align the bite properly.

c. Hand and Foot Surgery (Syndactyly Release)

- Goal: To separate fused fingers and toes, improving function.
- Procedure: Performed in stages, usually starting between 6-24 months of age.
- Outcome: Helps improve hand movement and grip, and hence development.

2. Supportive Therapies

Given Apert syndrome can affect speech, learning, and mobility, additional therapies may be needed:

- Speech Therapy: to help with speech delays or difficulties caused by facial structure differences.
- Physiotherapy & Occupational Therapy: improves motor skills and hand function.
- Hearing and Vision Care: regular check-ups with an audiologist and ophthalmologist to address hearing loss or vision problems.
- Dental and Orthodontic Care: needed to address misaligned teeth and jaw development issues.

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3. Long-Term Care and Monitoring (as required)

- Neurological assessments to monitor brain growth and detect issues like hydrocephalus (i.e., fluid build-up in the brain).
- Sleep studies to check for breathing problems like obstructive sleep apnoea.
- Psychological support & special education to address learning difficulties and social challenges.

4. Prenatal and Genetic Counselling

- If parents are concerned about passing Apert syndrome to their children, genetic counselling can help assess risks.

What next?

We are here to help. Explore support services on our website by scanning the QR code below. You can also explore more information on our online Resource Hub.

Did you know?

Apert syndrome was first described by a French physician named Eugène Apert in 1906.

In Australia, approximately 1 baby per 100,000 births was diagnosed with Apert syndrome between 1983 and 2010.*

References:

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How we can support you:

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