CONDITION

Apert syndrome

Learn how Apert syndrome affects the skull, face, hands, feet, breathing, hearing, and development, and how coordinated specialist care can help.

WHAT IT IS
Rare genetic condition affecting skull and limb development

BODY SYSTEM
Bones and joints

MAIN SYMPTOMS
Early skull closure, joined fingers and toes, facial differences, breathing problems

WHAT CAUSES IT
Inherited or new FGFR2 variant

HOW IT IS DIAGNOSED
FGFR2 genetic testing

MAIN TREATMENT
Specialist care and reconstructive surgery

CAN IT BE CURED?
No, genetic condition requiring lifelong care

IS IT AN EMERGENCY?
Sometimes, with severe trouble breathing

LONG-TERM OUTLOOK
Effects vary widely and require lifelong follow-up

Overview

Apert syndrome is a rare genetic condition that affects the development of the skull, face, hands, and feet. It is present from birth. The condition can also affect breathing, hearing, vision, teeth, speech, and learning.

In a baby's skull, several bones are separated by flexible joints called sutures. These joints allow the skull to expand as the brain grows. In Apert syndrome, one or more sutures close too early. This is called craniosynostosis . Early closure can change the shape of the head and may limit space for the growing brain.

Children with Apert syndrome also have syndactyly , meaning that fingers or toes are joined together. The bones, skin, or both may be connected. Hand differences can affect grasping and daily activities, while foot differences may affect shoe fit or walking.

The effects vary from one person to another. Some children have significant breathing or pressure-related problems that need early treatment. Others have fewer medical concerns. Intellectual ability and development also vary widely.

Care usually involves a craniofacial team rather than one clinician. This team may include specialists in neurosurgery, plastic and reconstructive surgery, ear, nose, and throat care, audiology, eye care, dentistry, genetics, hand surgery, speech therapy, and child development. Treatment is planned over time as the child grows.

Symptoms

The features of Apert syndrome are often recognizable at birth. The skull may be short from front to back and wider than expected. The forehead may appear tall, and the middle part of the face may not project forward as much as usual. This is called midface hypoplasia .

Common features can include:

Shallow eye sockets may leave the eyes more exposed. This can cause dryness, irritation, or difficulty closing the eyelids completely. Eye alignment problems, such as crossed or wandering eyes, can also occur.

Early skull fusion can sometimes lead to increased pressure around the brain. Signs may include persistent headache, vomiting, unusual sleepiness, irritability, worsening vision, or a decline in school performance. In babies, changes may be harder to recognize and can include poor feeding or an abnormal increase in head size.

Breathing problems may result from narrow nasal passages, a small midface, enlarged tonsils or adenoids, or narrowing lower in the airway. Obstructive sleep apnea occurs when breathing repeatedly becomes reduced or stops during sleep.

- Early closure of skull sutures
- Joined fingers and toes, often involving several digits
- Widely spaced or prominent eyes
- A small or flattened midface
- A small upper jaw and crowded teeth
- A high or narrow roof of the mouth, sometimes with a cleft palate
- Noisy breathing, snoring, or pauses in breathing during sleep
- Repeated middle-ear infections or hearing loss

- Speech or language difficulties
- Delayed development or learning differences
- Oily skin or severe acne, especially during adolescence

Causes and risk factors

Apert syndrome is caused by a disease-causing change, also called a pathogenic variant, in the FGFR2 gene. This gene provides instructions for a protein involved in cell growth, bone development, and the normal formation of skull sutures.

The condition follows an autosomal dominant inheritance pattern. This means one altered copy of the gene is enough to cause Apert syndrome.

Most affected children are born to parents who do not have the condition. In these cases, the genetic change occurred as a new event in the egg, sperm, or early embryo. Older paternal age is associated with a higher chance of new FGFR2 variants, but Apert syndrome can occur at any parental age. Nothing a parent did during pregnancy causes the condition.

A person with Apert syndrome has a chance of passing the altered gene to each child. A genetics professional can explain inheritance, testing options, and family-planning choices without directing a family toward a particular decision.

Diagnosis

A clinician may suspect Apert syndrome after examining a newborn's head shape, facial features, hands, and feet. The pattern of craniosynostosis and syndactyly helps distinguish it from other conditions that affect skull growth.

Genetic testing can confirm the diagnosis by identifying a pathogenic variant in FGFR2. Testing may also help clarify the diagnosis when physical findings overlap with another craniosynostosis syndrome.

The initial evaluation looks beyond the skull and limbs. Depending on the child's needs, it may include:

Computed tomography, or CT, can provide detailed images of the skull. Because CT uses radiation, the craniofacial team considers whether it is necessary and how to limit exposure. Other tests, including magnetic resonance imaging, may be used when clinicians need to evaluate the brain or fluid spaces.

Apert syndrome may sometimes be suspected before birth through ultrasound findings.

Prenatal genetic testing can confirm the diagnosis when a specific FGFR2 variant is identified. Families considering testing can meet with a genetic counselor to discuss what the results may and may not show.

- An eye examination, including checks of the optic nerves and ability to close the eyelids
- Hearing tests and an examination for middle-ear fluid
- Assessment of breathing while awake and asleep
- Feeding, speech, and developmental evaluations
- Dental and palate examinations
- Imaging of the skull and brain when it will guide treatment
- Evaluation of hand and foot bones before reconstructive planning

Treatment and management

There is no single treatment plan for every person with Apert syndrome. Care focuses on protecting the brain, eyes, hearing, airway, and development while improving hand function, dental alignment, and facial balance.

Cranial surgery is commonly performed during infancy or early childhood to expand and reshape the skull. Its goals may include creating room for brain growth, reducing the risk of increased pressure, and improving skull shape. The operation and timing depend on which sutures are fused, the child's age, symptoms, and the craniofacial center's approach. Some people need more than one skull procedure as they grow.

Midface advancement surgery moves the central facial bones forward. It may improve breathing, protect the eyes, and help facial and dental alignment. Timing is individualized. Significant airway or eye problems may lead to earlier treatment, while other procedures may be planned later in childhood or adolescence.

Hand surgery can separate selected fingers and improve useful grasp. More than one operation may be needed because the pattern of bone and skin connection is often complex. The goal is practical hand function rather than creating fingers that look or move exactly like typical fingers. Occupational therapy can help with strength, coordination, adaptive tools, and daily tasks.

Other management may include:

Families may benefit from psychological support and contact with craniofacial peer groups. Children and adults can face unwanted attention, questions, or teasing because of visible

differences. Support should respect the person's identity and preferences rather than treating appearance alone as a medical problem.

- Hearing aids or ear tubes when hearing or middle-ear problems are present
- Treatment for sleep apnea or other airway obstruction
- Lubricating eye treatments or procedures to protect exposed eyes
- Orthodontic and dental care for crowded teeth and jaw differences
- Speech or feeding therapy
- Physical and occupational therapy
- Educational assessment and individualized school support
- Standard treatment for acne or other skin concerns

Complications

Craniosynostosis can contribute to increased pressure inside the skull. Without recognition and treatment, prolonged pressure may affect vision or brain function. Pressure can recur even after surgery, so continued monitoring is important.

Shallow eye sockets can expose the surface of the eye and increase the risk of corneal injury. Misaligned eyes or unequal vision can interfere with normal visual development in children.

Narrow airways may lead to sleep apnea, disturbed sleep, poor concentration, daytime tiredness, or stress on the heart and lungs in severe cases. Hearing loss may be temporary or lasting. When hearing problems are not addressed, they can affect speech, language, and learning.

Crowded teeth, an underdeveloped upper jaw, and palate differences can make chewing, cleaning the teeth, and speaking more difficult. Joined fingers may limit fine motor skills. Foot structure can cause pressure areas or problems finding comfortable shoes.

Surgery has risks such as bleeding, infection, scarring, or the need for another procedure. The surgical team should explain the expected benefits, alternatives, and recovery before each operation.

Seek emergency care for severe trouble breathing, blue or gray lips, a seizure, loss of consciousness, or sudden major changes in alertness. Urgent medical assessment is also appropriate for repeated vomiting with headache, sudden vision changes, new inability to close an eye, severe eye pain, or rapidly worsening sleep-related breathing.

Follow up

Apert syndrome requires long-term follow-up because medical needs change with growth. Routine visits with a craniofacial team allow specialists to coordinate testing and avoid unnecessary duplication.

Follow-up commonly includes checks of skull growth, head shape, vision, hearing, breathing, sleep, dental development, hand function, speech, and learning. Clinicians may examine the optic nerves for signs of pressure even when a child does not report headaches. Sleep testing may be recommended when there is persistent snoring, pauses in breathing, restless sleep, or daytime symptoms.

Developmental progress should be reviewed during early childhood and school years. Support can be adjusted when new academic, communication, movement, or social needs appear. Transition planning can help adolescents move from pediatric to adult medical, dental, and mental health services.

Contact the care team promptly about new headaches, repeated vomiting, worsening snoring, breathing pauses, reduced hearing, eye irritation, changes in vision, loss of skills, or a clear change in behavior or school performance. These symptoms do not always signal a serious problem, but they should not wait for a distant routine appointment.

Sources

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