

Apert syndrome

Description

Apert syndrome is a genetic disorder characterized by skeletal abnormalities. A key feature of Apert syndrome is the premature closure of the bones of the skull (craniosynostosis). This early fusion prevents the skull from growing normally and affects the shape of the head and face. In addition, a varied number of fingers and toes are fused together (syndactyly).

Craniosynostosis causes many of the characteristic facial features of Apert syndrome. Premature fusion of the skull bones prevents the head from growing normally, which leads to a sunken appearance in the middle of the face (midface hypoplasia), a beaked nose, a wrinkled forehead, and an opening in the roof of the mouth (a cleft palate). In individuals with Apert syndrome, an underdeveloped upper jaw can lead to dental problems, such as missing teeth, irregular tooth enamel, and crowded teeth.

Many individuals with Apert syndrome have vision problems due to eye abnormalities, which can include bulging eyes (exophthalmos), wide-set eyes (hypertelorism), outside corners of the eyes that point downward (downslanting palpebral fissures), eyes that do not look in the same direction (strabismus), and shallow eye sockets (ocular proptosis). Some people with Apert syndrome have hearing loss or recurrent ear infections due to malformed ear structures.

Abnormal development of structures in the face and head can also cause partial blockage of the airways and lead to breathing difficulties in people with Apert syndrome. Craniosynostosis also affects development of the brain, which can disrupt intellectual development. Cognitive abilities in people with Apert syndrome range from normal to mild or moderate intellectual disability.

Individuals with Apert syndrome have syndactyly of the fingers and toes. The severity of the fusion varies, although the hands tend to be more severely affected than the feet. Most commonly, three digits on each hand and foot are fused together. In the most severe cases, all of the fingers and toes are fused. Rarely, people with Apert syndrome may have extra fingers or toes (polydactyly). Some people with Apert syndrome have abnormalities in the bones of the elbows or shoulders. These bone problems can restrict movement and impede everyday activities. In some people, abnormalities occur in both sides of the body, but in others, only one side is affected.

Additional signs and symptoms of Apert syndrome can include unusually heavy

sweating (hyperhidrosis), oily skin with severe acne, or patches of missing hair in the eyebrows.

Frequency

Apert syndrome affects an estimated 1 in 65,000 to 88,000 newborns.

Although parents of all ages can have a child with Apert syndrome, the risk is increased in older fathers.

Causes

Mutations in a gene known as *FGFR2* cause Apert syndrome. This gene provides instructions for making a protein called fibroblast growth factor receptor 2 (FGFR2). Among its multiple functions, the FGFR2 protein plays a key role in development before birth by signaling immature cells to become bone cells. A mutation in a specific part of the *FGFR2* gene alters the protein, increasing its signaling. The abnormal signaling causes the cell to mature too quickly and promotes the premature fusion of bones in the skull, hands, and feet.

[Learn more about the gene associated with Apert syndrome](#)

- FGFR2

Inheritance

Apert syndrome is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. Nearly all cases of this condition result from new (de novo) mutations in the gene that occur during the formation of reproductive cells (eggs or sperm) in an affected individual's parent or in early embryonic development. These cases occur in people with no history of the disorder in their family.

Other Names for This Condition

- Acrocephalosyndactyly
- Acrocephalosyndactyly type I
- Apert's syndrome
- Type I acrocephalosyndactyly

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Acrocephalosyndactyly type I (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0001193/>)

Genetic and Rare Diseases Information Center

- Apert syndrome (<https://rarediseases.info.nih.gov/diseases/5833/index>)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

Clinical Trials

- ClinicalTrials.gov (<https://clinicaltrials.gov/search?cond=%22Apert syndrome%22>)

Catalog of Genes and Diseases from OMIM

- APERT SYNDROME (<https://omim.org/entry/101200>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28apert+syndrome%5BTIAB%5D%29+OR+%28acrocephalosyndactyly%5BTIAB%5D%29+OR+%28acrocephaly%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D>)

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