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Apert Syndrome

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ABSTRACT

Apert syndrome is a genetic disorder characterized by skeletal abnormalities. 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. 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). The abnormal signaling causes the cell to mature too quickly and promotes the premature fusion of bones in the skull, hands, and feet. 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. Genetic testing can usually identify Apert syndrome or another cause of abnormal skull formation. Apert syndrome has no known cure. Surgery to correct the abnormal connections between bones is the main treatment for Apert syndrome.

Key words: Apert syndrome, Fibroblast growth factor, Craniosynostosis, FGFR2

DEFINITION

Apert syndrome, also called acrocephalosyndactyly, is a genetic syndrome characterized by anomalies of the skull, face and limbs. Gene mutations are responsible for causing the early fusion of the skull, hand and feet bones.

INCIDENCE

Apert syndrome is estimated to affect 1 in 65,000 babies individuals worldwide.

CAUSE

A genetic mutation of a gene, fibroblast growth factor receptor-2 or FGFR2, which is responsible for skeletal development causes Apert syndrome. When the gene mutation occurs, receptors don't communicate with fibroblast growth factors causing joints (sutures) between bones to close too soon during fetal development. The irregular formation of the bones leads to malformations of their body.

PATHOGENESIS

Apert syndrome inherited with point mutations of either Ser252Trp or Pro253Arg in fibroblast growth factor receptor 2 (FGFR2) on chromosome 10q25. FGFR2 belongs to a complex system of intracellular signalling consisting of multiple fibroblast growth factors (FGFs) and their receptors FGFRs. This signalling network functions in the control of cell proliferation, differentiation, migration. The FGFR2 is active at the metaphysis; diaphysis and also in the interdigital mesenchyme. Mutations in the FGFR2 gene cause prolonged signalling, which can promote the premature fusion of bones in the skull, hands, and feet.

SIGNS AND SYMPTOMS

Craniosynostosis: early closure of one or more of the seams between the skull bones causing an abnormal skull. This results in a skull shape with increased vertical height.

Midface hypoplasia: decreased growth of the midface. This causes a crescent moon or sunken facial appearance with depressed nasal bridge and beak nose. Decreased growth of the central face can contribute obstructive sleep apnea and airway concerns.

Turribrachycephaly a cone-shaped skull

Syndactyly: fusion of the fingers and toes. The hand can resemble a spade, mitten or rosebud.

Other common characteristics include large elongated forehead, shallow eye sockets causing prominent eyes, narrow palate with or without a cleft, fused bones in the arms, elbows and hips, and hearing loss. The characteristic feature that distinguishes Apert syndrome from other types of syndromic craniosynostosis is the presence of hand anomalies, most commonly fused or webbed fingers (syndactyly).

DIAGNOSIS

An early diagnosis is possible during pregnancy with a prenatal 2D or 3D ultrasound or MRI to track baby's skeletal development.

Skull radiograph or CT scan of the head to determine the nature of the bone abnormalities.

Apert syndrome can often be diagnosed at birth or at an early age. Genetic testing can usually identify Apert syndrome or another cause of abnormal skull formation. Genetic testing, looking for a mutation of the FGFR2 gene, to confirm the diagnosis.

TREATMENT

Treatment for Apert syndrome varies based on the severity of child's diagnosis. Treatment most often involves a type of surgery to alleviate symptoms. Symptoms that affect their skull or brain (craniosynostosis or hydrocephalus), healthcare provider will schedule surgery between two to four months after they are born to correct the condition by inserting a tube (shunt) to drain the fluid and release pressure from the brain. Reconstructive or corrective surgery could adjust any part of child's body that formed abnormally.

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