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My46 Trait Profile

Apert syndrome

Other Names: Acrocephalosyndactyly type II

Apert syndrome is a rare genetic condition caused be early closure of one or more skull bones. It is caused by mutations in the FGFR2 gene which makes the protein called fibroblast growth factor receptor 2.

Characteristics of Apert syndrome

Apert syndrome is one of a group of related conditions that result in craniosynostosis (premature or early fusion of some of the plates of bone forming the skull) causing a tall, broad forehead, wide spacing and prominence of the eyes, as well as a flattened midface. Many complications can result from these changes including: hearing loss, narrow or cleft palate, crowding of the teeth, and breathing problems. The hands and feet of individuals with Apert are often affected with variable fusion of the bones and/or skin. Other bone problems occur at times such as fusion of the bones of the forearm resulting in limited ability to turn the hands. Intelligence is affected in about half of individuals with Apert syndrome ranging from mild learning disabilities to more significant intellectual disability.

Diagnosis/Testing

Most individuals with Apert syndrome have a change or mutation in a gene called FGFR2. This gene is responsible for regulating cell growth, specialization and delivery, all of which are important in limb development, as well as skull bone formation and fusion. Mutations in the FGFR2 gene result in early closure of the bones of the skull and in problems in the formation and separation of the skin and bones of the fingers and toes.

Management/Surveillance

Management of Apert syndrome often includes brain imaging, and x-ray evaluations of the limbs and spine. Individuals with Apert syndrome are often followed by eye doctors (ophthalmology), audiologists, and bone doctors (orthopedics). Individuals with Apert syndrome often undergo several surgeries to reshape the skull. Involvement of early intervention services to monitor for developmental concerns and monitoring by the craniofacial team is recommended.

Mode of inheritance

Apert syndrome is inherited in an autosomal dominant pattern. This means inheriting one FGFR2 mutation is enough for an individual to be affected and show signs of Apert syndrome. The mutation can be inherited from an affected parent or it can occur brand new (de novo) in an affected child.

Risk to family members

The risk to family members depends on whether or not the individual with Apert syndrome has a parent affected with the condition. If a parent also has Apert syndrome, the risk of their having another child with Apert syndrome is 50% with each pregnancy. If a parent does not have Apert syndrome, the risk of other siblings being affected is very low.

Special considerations

None

Resources

AboutFace
http://aboutface.ca
Ameriface
http://www.ameriface.org/
Children's Craniofacial Association
http://www.ccakids.com/
Genetics Home Reference: Apert syndrome
http://ghr.nlm.nih.gov/condition/apert-syndrome

References

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Robin NH, Falk MJ, Halderman-Englert CR. (Updated 7 June 2011). FGFR-Related Craniosynostosis Syndromes. In: GeneReviews at GeneTests Medical Genetics Information Resource (database online). Copyright, University of Washington, Seattle. 1997-2013. Available at http://www.ncbi.nlm.nih.gov/books/NBK1455/. Accessed [09/11/2013].

Created: 09/2013 **Created by:** Heather Stalker, MSc, CGC
Updated: mm/yyyy **Edited by:** Seema Jamal, MSc, LCGC