Adams Oliver syndrome

Other Names: Aplasia cutis congenita with terminal transverse limb defects

Adams-Oliver syndrome is a genetic condition characterized by birth defects involving the scalp and limbs. It is caused by mutations in the genes ARHGAP31, DOCK6, and RBPJ, each of which makes a protein involved in human development.

Characteristics of Adams Oliver syndrome

Adams-Oliver syndrome is a rare genetic condition characterized by aplasia cutis congenita (absence of the skin usually appearing as hairless scarred areas on the back of the scalp) in combination with terminal transverse limb defects (abnormalities of the fingers, toes, arms, and/or legs). The limb defects seen in this condition can include short fingers and/or toes, missing terminal phalanges (the tips of fingers or toes), and missing entire hands or feet. Additional features may include congenital heart defects and vascular anomalies (problems involving the blood vessels). Some individuals with AOS may also have pulmonary hypertension (high blood pressure in the arteries to the lungs). The signs and symptoms of the AOS are highly variable within families as well as between other unrelated, affected individuals. Intelligence is usually normal in individuals with AOS, although intellectual disability has been described in some individuals.

Diagnosis/Testing

Most individuals with AOS have a change or mutation in a gene called ARHGAP31. This gene controls two proteins that play an important role in growth, division, and movement of cells. Individuals with an ARHGAP31 gene mutation have AOS type 1. AOS type 2 and AOS type 3 are less common and are caused by mutations in the DOCK6 gene and the RBPJ gene, respectively.

Management/Surveillance

Management of AOS depends on the severity of the clinical features. Some individuals with AOS need surgical treatment of their scalp defects by skin grafting, and surgery to repair their heart defects. Depending on the presentation of the limb anomalies, modifications to living space may be needed.

Mode of inheritance

AOS may be inherited in one of two patterns of inheritance: autosomal dominant and autosomal recessive. Autosomal dominant inheritance:

AOS type 1 and AOS type 3 are inherited in an autosomal dominant pattern. This means inheriting one mutation is enough for an individual to be affected and show signs of AOS type 1 or AOS type 3. The mutation can be inherited from an affected parent or it can occur brand new (de novo) in an affected child.

Autosomal recessive inheritance:

AOS type 2 is inherited in an autosomal recessive pattern. This means that an individual has to inherit two DOCK6 mutations (i.e., one from each parent) to be affected with AOS type 2. If both parents are carriers of a DOCK6 mutation, they have a 1 in 4 (25%) chance with each pregnancy of having a child with AOS type 2.
**Risk to family members**

The risk to family members depends on the pattern of inheritance.

**Autosomal dominant inheritance:**

The risk to family members depends on whether or not the individual with AOS type 1 or AOS type 3 has a parent affected with AOS. If a parent also has the condition, the risk of having a child with AOS type 1 or AOS type 3 is 50% with each pregnancy. If a parent does not have the condition, the risk of other siblings being affected is very low.

**Autosomal recessive inheritance:**

Parents of a child with AOS type 2 are carriers of AOS type 2. If a sibling of a child with AOS type 2 is unaffected, he/she has a 2 in 3 (66%) chance of being a carrier of AOS type 2.

**Special considerations**

None

**Resources**

AOSupport
http://www.aosupport.org/

Genetics Home Reference: ARHGAP31

**References**


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