Adams-Oliver Syndrome 2

Alternative Names
AOS2

Record Category
Disease phenotype

WHO-ICD
Congenital malformations, deformations and chromosomal abnormalities> Other congenital malformations.

Incidence per 100,000 Live Births
Unknown

OMIM Number
614219

Mode of Inheritance
Autosomal recessive

Gene Map Locus
19p13.2

Description

Adams-Oliver Syndrome (AOS) is a rare genetic disease characterized by aplasia cutis congenita (ACC) and terminal transverse limb defects (TTLD) with varying degrees of severity in each individual. Some patients may show very mild clinical features, while others may be severe. The prevalence of AOS is unknown. Variable clinical features are observed in different individuals, including malformations of the hands, arms, feet and/or legs that range from hypoplastic fingers and toes to absent hands and/or lower legs, and occasionally show intellectual deficit. Physical anomalies may present in some AOS patients including congenital cataract, strabismus and microphthalmia, congenital heart malformations, and hepatoportal sclerosis. Extensive lethal anomalies are also possible.

The diagnosis is suspected at birth based on clinical findings of ACC of the scalp and TTLD. The treatment options for Adams Oliver Syndrome are supportive with surgical management of the defects. The limb and scalp defects require orthopedic treatment.

Molecular Genetics
Adams-Oliver Syndrome (AOS) is transmitted as autosomal dominant and autosomal recessive modes of inheritance. Mutations in the DOCK6 gene, located on 19p13.2 chromosome, are the cause of the autosomal recessive form of ASO2. The protein encoded by DOCK6 gene is involved in the regulation of proteins called GTPases, which transmit signals that are critical for various aspects of embryonic development of the limbs, skull, and heart.

Epidemiology in the Arab World

Saudi Arabia
Shaheen et al. (2011) described two unrelated individuals born to consanguineous parents with autosomal recessive Adams-Oliver syndrome. The first patient was an 11-months-old girl who presented with severe and global developmental delay, recurrent seizures, and poor vision. She had microcephaly, large cutis aplasia of the scalp, optic atrophy, distal reduction of the fingers and toes bilaterally and an absence of distal phalanges and nails, and axial hypotonia with appendicular hypertonia. The second patient was a 3.5-year-old girl who had congenital deformity of the hands and feet. At birth, she presented with defects of her hands and feet and cutis aplasia of the scalp. Her finger and toe nails were either absent or severely hypoplastic, as were the distal phalanges, and her hands appeared stubby with distorted creases. Both patients were found to carry separate homozygous mutations in the DOCK6 gene.

References
Shaheen R, Faqeih E, Sunker A, Morsy H, Al-Sheddi T, Shamseldin HE, Adly N, Hashem M, Alkuraya FS. Recessive mutations in DOCK6, encoding the guanidine nucleotide exchange factor DOCK6, lead to abnormal actin cytoskeleton organization and

Adams-Oliver syndrome. Am J Hum Genet. 2011 12; 89(2):328-33. PMID: 21820096

Related CTGA Records

Dedicator of Cytokinesis 6

External Links

https://ghr.nlm.nih.gov/condition/adams-oliver-syndrome

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Expert=974

http://www.webmd.com/children/adams-oliver-syndrome

Contributors

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