Adams-Oliver syndrome

Adams-Oliver syndrome is a very rare syndrome, first described in 1945 by Forrest H. Adams and CP Oliver [1]. The main manifestatins of the syndrome are **aplasia cutis congenita** (congenital defect of the skin, especially in the hairy part of the head) and **transverse defects of the limbs** (especially the lower). There are also defects of other organs such as congenital heart defects, microphtalmos, cryptorchidism.

The syndrome is **genetically heterogenous**, inheritance is most often autosomal dominant, amd sporadically appearing cases of autosomal recessive cases are describe. In 2011, a possible association of this syndrome with the mutations gene *ARHGAP31* (3q13.3) [2].

Links

References

1. ENERSEN, Ole Daniel. *Adams-Oliver syndrome (WhoNamedIt)* [online]. [cit. 2011-10-22]. <http://www.whonamedit.com/synd.cfm/717.html>.
2. SOUTHGATE, Laura – MACHADO, Katie M – SNAPE,. Gain-of-function mutations of ARHGAP31, a Cdc42/Rac1 GTPase regulator, cause syndromic cutis aplasia and limb anomalies. *Am J Hum Genet* [online]. 2011, y. 5, p. 574-85, Available from <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3146732/?tool=pubmed>. ISSN 0002-9297 (print), 1537-6605.

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