Rombo syndrome

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Published in Atlas Database: September 2011
Online updated version: http://AtlasGeneticsOncology.org/Kprones/RomboID10169.html
DOI: 10.4267/2042/46949

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Identity

Inheritance
Rare disorder, with less than 10 cases described, with a probable autosomal dominant transmission, as suggested by the family tree of four generations in the princeps report (Michaëlsson et al., 1981).

Differential Diagnosis
Resembles Bazex-Dupré-Christol syndrome, which is a X-linked dominant disease.

Neoplastic risk
Basal cell carcinomas are a frequent complication.

Clinics

Phenotype and clinics
Skin changes appear at the age of 6-10 years, with cyanotic redness, acral erythema, thin implantation of hair and absent eyelashes (hypotrichosis). Atrophoderma vermiculatum (severe skin atrophy) of the face and sun-exposed areas, telangiectasia and milia-like papules develop in adulthood. Histology of the skin shows highly irregular distribution of elastin in the upper dermis, with areas without elastin and others with clumps of elastin, vascular proliferation and lymphocytes infiltration (Michaëlsson et al., 1981; Van Steensel et al., 2001).

Note
The gene involved in this rare disease is unknown.

References


This article should be referenced as such: