Opis choroby *

Definicja

Hallermann-Streiff syndrome is a rare genetic syndrome characterized mainly by head and facial abnormalities such as bird-like facies (with beak-shaped nose and retrognathia), hypoplastic mandible, brachycephaly with frontal bossing, dental abnormalities (e.g. absence of teeth, natal teeth, supernumerary teeth, severe agenesis of permanent teeth, enamel hypoplasia) hypotrichosis, various ophthalmic disorders (e.g. congenital cataracts, bilateral microphthalmia, ptosis, nystagmus) and atrophy of skin (especially around the center of face and nose) as well as telangiectasia and proportionate short stature. Intellectual disability is reported in some cases.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
François dyscephalic syndrome
Zespół dyscefalii Françoisa
Oculomandibulofacial syndrome

Kod ORPHA
2108

Kod OMIM
234100

Kod ICD10
Q87.0

Kod ICD11
LD2B

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*Źródło

orphanet