

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	Synonimy
Zespół wad wrodzonych François	dyscephalic syndrome
	Zespół dyscefalii François
	Oculomandibulofacial syndrome

Kod ORPHA	Kod OMIM	Kod ICD10
2108	234100	Q87.0

Kod ICD11
LD2B

*Źródło

orphanet