

Opis choroby *

Definicja

A rare frontonasal dysplasia malformation syndrome characterized by an oxycephalic skull with craniosynostosis, wide nose with anteverted nostrils, hirsutism at base of nose, agenesis of the nasolacrimal ducts, and bilateral, symmetrical nasolabial cysts on upper lip. Additional features may include hypertelorism. There have been no further descriptions in the literature since 1991.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

157832

Kod OMIM

123050

Kod ICD10

Q30.8

Kod ICD11

LA70.Y

*Źródło

orphanet