Opis choroby *

Definicja

A rare frontonasal dysplasia characterized by a craniofacial phenotype comprising frontal bossing with high anterior hairline, ptosis, hypertelorism, epicanthus inversus, flat nasal bridge, and broad nasal tip. Large anterior fontanelle, sagittal synostosis, and cranial base anomalies have also been described.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
SIX2-related FND
SIX2-related FND

Kod ORPHA
488437

Kod OMIM
-

Kod ICD10
Q75.8

Kod ICD11
-

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*Źródło

orphanet