Opis choroby *

Definicja

Craniosynostosis, Herrmann-Opitz type is a rare bone development disorder characterized by intellectual disability, short stature, turribrachycephaly, facial dysmorphism (i.e. severe hypertelorism, hypoplasia of supraorbital ridges, abnormal ears, and micrognathia), bony defects of the occiput, and digital anomalies (incl. syndactyly, oligodactyly, and/or brachydactyly). Urethral atresia has also been reported. There have been no further descriptions in the literature since 1987.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
2145

Kod OMIM
-

Kod ICD10
Q75.0

Kod ICD11
LD24.GY

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*Źródło

orphanet