

Opis choroby *

Definicja

Trigonocephaly-broad thumbs syndrome is characterized by neonatal trigonocephaly and multiple anomalies including craniosynostosis, shallow orbits, unusual nose, deviation of the terminal phalanges of fingers 1, 2, and 5, and broad toes with duplication of the terminal phalanx. It has been described in a mother and her son. It is transmitted as an autosomal dominant trait.

Dane

Klasyfikacja

Zespół wad wrodzonych Hunter-Rudd-Hoffmann syndrome

Synonimy

Zespół Huntera, Rudda i Hoffmanna

Kod ORPHA

3365

Kod OMIM

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Kod ICD10

Q87.0

Kod ICD11

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*Źródło

orphanet