Opis choroby *

Definicja

Craniosynostosis-intracranial calcifications syndrome is a form of syndromic craniosynostosis characterized by pancraniosynostosis, head circumference below the mid-parental head circumference, mild facial dysmorphism (prominent supraorbital ridges, mild proptosis and maxillary hypoplasia) and calcification of the basal ganglia. The disease is associated with a favorable neurological outcome, normal intelligence and is inherited in an autosomal recessive manner.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Longman-Tolmie syndrome
Zespół Longmana i Tolmiego

Kod ORPHA
52054

Kod OMIM
608432

Kod ICD10
Q87.0

Kod ICD11
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*Źródło

orphanet