Opis choroby *

Definicja

Hunter-McAlpine craniosynostosis is characterised by craniosynostosis, intellectual deficit, short stature, facial dysmorphism (oval face with almond-shaped palpebral fissures, droopy eyelids and a small nose) and minor distal anomalies. It has been described in 10 patients. Transmission is autosomal dominant and the syndrome is associated with partial duplication of the long arm of chromosome 5 (5q35-5qter).

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Zespół Hunter-McAlpine

Kod ORPHA
97340

Kod OMIM
601379

Kod ICD10
Q87.0

Kod ICD11
LD24.GY

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*Źródło

orphanet