

## 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

#### Synonimy

Zespół wad wrodzonych Zespół Hunter-McAlpine

#### Kod ORPHA

97340

#### Kod OMIM

601379

#### Kod ICD10

Q87.0

#### Kod ICD11

LD24.GY

---

#### \*Źródło

orphanet