## Opis choroby *

Definicja

A rare neurocutaneous syndrome characterized by the association of cerebellum (rhombencephalosynapsis), cranial nerves (trigeminal anesthesia), and scalp (alopecia) abnormalities. Other features observed in patients were craniosynostosis, midfacial hypoplasia, bilateral corneal opacities, low-set ears, short stature, moderate intellectual impairment and ataxia. Hyperactivity, depression, self-injurious behaviour and bipolar disorder have also been reported.

Dane

**Klasyfikacja**
Zespół wad wrodzonych

Synonimy
Cerebellotrigeminal-dermal dysplasia syndrome
Dysplazja móżdżkowo-trójdzielno-skórna
Kraniosynostoza - łysienie - defekt mózgu
Craniosynostosis-alopecia-brain defect syndrome

**Kod ORPHA**
1532

**Kod OMIM**
601853

**Kod ICD10**
Q07.8

**Kod ICD11**
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*Źródło

orphanet