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
-

---

*Źródło

orphanet