Opis choroby *

Definicja

Biemond syndrome type 2 (BS2) is a rare genetic neurological and developmental disorder reported in a very small number of patients with a poorly defined phenotype which includes iris coloboma, short stature, obesity, hypogonadism, postaxial polydactyly, and intellectual disability. Hydrocephalus and facial dysostosis were also reported. BS2 shares features with Bardet-Biedl syndrome. There have been no further descriptions in the literature since 1997.

Dane

Klasyfikacja
Choroba

Synonimy
Hypogonadism-short stature-coloboma-preaxial polydactyly syndrome
Zespół hipogonadyzm-niski wzrost-szczelina-polidaktylia przedosiowa

Kod ORPHA
141333

Kod OMIM
210350

Kod ICD10
Q87.8

Kod ICD11
-

---

*Źródło

orphanet