Opis choroby *

Definicja

Bonnemann-Meinecke-Reich syndrome is a syndrome of multiple congenital anomalies characterized by an encephalopathy which predominantly occurs in the first year of life and presenting as psychomotor delay. Additional features of the disease include moderate dysmorphia, craniosynostosis, dwarfism (due to growth hormone deficiency), intellectual disability, spasticity, ataxia, retinal degeneration, and adrenal and uterine hypoplasia. The disease has been described in only two families, with each family having two affected siblings. An autosomal recessive inheritance has been suggested. There have been no further descriptions in the literature since 1991.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Encephalopathy-intracerebral calcification-retinal degeneration syndrome
Encefalopatia - zwapnienia wewnątrzczaszkowe - degeneracja siatkówki

Kod ORPHA
1261

Kod OMIM
225755

Kod ICD10
Q04.8

Kod ICD11
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*Źródło

orphanet