Opis choroby *

Definicja

Grubben-de Cock-Borghgraef syndrome is a rare intellectual disability syndrome characterized by pre- and postnatal growth deficiency, generalized muscular hypotonia, developmental delay (particularly of speech and language), hypotrophy of distal extremities, small and puffy hands and feet, eczematous skin and dental anomalies (i.e. small, widely-spaced teeth). Partial agenesis of the corpus callosum and a selective immunoglobulin IgG2 subclass deficiency have also been reported in some patients.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Developmental delay-hypotonia-extremities hypertrophy syndrome
Opóźnienie rozwoju - hipotonia - hipertrofia kończyn

Kod ORPHA
2101

Kod OMIM
233810

Kod ICD10
Q87.8

Kod ICD11
-

---

*Źródło

orphanet