

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

Kod ORPHA	Kod OMIM	Kod ICD10
2101	233810	Q87.8

Kod ICD11

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*Źródło

orphanet