Grubben-de Cock-Borghgraef syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Grubben-de Cock-Borghgraef syndrome. ORPHA:2101

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.