Microcephalic primordial dwarfism, Toriello type

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Microcephalic primordial dwarfism, Toriello type. ORPHA:2643

Microcephalic primordial dwarfism, Toriello type is characterised by growth retardation with prenatal onset, cataracts, microcephaly, intellectual deficit, immune deficiency, delayed ossification and enamel hypoplasia. It has been described in two siblings. Transmission is autosomal recessive.