Marfanoid habitus-autosomal recessive intellectual disability syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Marfanoid habitus-autosomal recessive intellectual disability syndrome. ORPHA:2463

Marfanoid habitus intellectual deficit, autosomal recessive is a very rare multiple congenital anomalies syndrome described in four sibs and characterized by intellectual deficit, flat face and some skeletelal features of Marfan syndrome (see this term) such as tall stature, dolichostenomelia, arm span larger than height, arachnodactyly of hands and feet, little subcutaneous fat, muscle hypotonia and intellectual deficit.