Osteoporosis-macrocephaly-blindness-joint hyperlaxity syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Osteoporosis-macrocephaly-blindness-joint hyperlaxity syndrome. ORPHA:2787

Osteoporosis-macrocephaly-blindness-joint hyperlaxity syndrome is characterised by osteoporosis, macrocephalus, brachytelephalangy, and hyperextensibility of the joints. Congenital amaurosis and intellectual deficit have also been reported. This syndrome has been described in three members of one family.