Osteopenia-myopia-hearing loss-intellectual disability-facial dysmorphism syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Osteopenia-myopia-hearing loss-intellectual disability-facial dysmorphism syndrome. ORPHA:91133

Osteopenia-myopia-hearing loss-intellectual disability-facial dysmorphism syndrome is characterised by severe hypertelorism, brachycephaly, abnormal ears, sloping shoulders, enamel hypoplasia, osteopaenia with frequent fractures, severe myopia, mild to moderate sensorineural hearing loss and mild intellectual deficit. It has been described in two brothers born to first-cousin parents. No chromosomal anomalies were detected. Transmission appears to be autosomal recessive or X-linked.