Congenital muscular dystrophy-infantile cataract-hypogonadism syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Congenital muscular dystrophy-infantile cataract-hypogonadism syndrome. ORPHA:1875

Congenital muscular dystrophy-infantile cataract-hypogonadism syndrome is characterized by congenital muscular dystrophy, infantile cataract and hypogonadism. It has been described in seven individuals from an isolated Norwegian village and in one unrelated individual. Transmission appears to be autosomal recessive.