X-linked intellectual disability-hypogammaglobulinemia-progressive neurological deterioration syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. X-linked intellectual disability-hypogammaglobulinemia-progressive neurological deterioration syndrome. ORPHA:85317

X-linked intellectual disability-hypogammaglobulinemia-progressive neurological deterioration syndrome is characterized by moderate intellectual deficit, bilateral single palmar creases, seizures, variable hypogammaglobulinemia and characteristic features (synophrys, prognathism, and hirsutism). It has been reported in three males from two generations of one family. All underwent progressive neurological deterioration. This syndrome is transmitted as an X-linked trait, and the causative gene is located between Xq21.33 and Xq23.