Deafness-intellectual disability syndrome, Martin-Probst type

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Deafness-intellectual disability syndrome, Martin-Probst type. ORPHA:85321

Deafness-intellectual disability syndrome, Martin-Probst type is characterised by severe bilateral deafness, intellectual deficit, umbilical hernia and abnormal dermatoglyphics. It has been described in three males from three generations of one family. Mild facial dysmorphism (telangiectasias, hypertelorism, dental anomalies and a wide nasal root) was also present. Short stature, pancytopaenia, microcephaly, and renal and genitourinary anomalies were present in some of the patients. The mode of transmission is X-linked recessive and the causative gene has been localised to the q1-21 region of the X chromosome.