X-linked intellectual disability-craniofacioskeletal syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. X-linked intellectual disability-craniofacioskeletal syndrome. ORPHA:163979

X-linked intellectual disability-craniofacioskeletal syndrome is a rare, hereditary, syndromic intellectual disability characterized by craniofacial and skeletal abnormalities in association with mild intellectual disability in females and early postnatal lethality in males. In addition to mild cognitive impairment, females present with microcephaly, short stature, skeletal features and extra temporal lobe gyrus. In males, intrauterine growth impairment, cardiac and urogenital anomalies have been reported.