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X-linked intellectual disability-cardiomegaly-congestive heart failure syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. X-linked intellectual disability-cardiomegaly-congestive heart failure syndrome. ORPHA:324410*

X-linked intellectual disability-cardiomegaly-congestive heart failure syndrome is a rare X-linked syndromic intellectual disability disorder characterized by profound intellectual disability, global developmental delay with absent speech, seizures, large joint contractures, abnormal position of thumbs and middle-age onset of cardiomegaly and atrioventricular valve abnormalities, resulting in subsequent congestive heart failure. Additional features include variable facial dysmorphism (notably large ears with overfolded helix) and large testes.