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X-linked myotubular myopathy-abnormal genitalia syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. X-linked myotubular myopathy-abnormal genitalia syndrome. ORPHA:456328*

X-linked myotubular myopathy-abnormal genitalia syndrome is a rare chromosomal anomaly, partial deletion of the long arm of chromosome X, characterized by a combination of clinical manifestations of X-linked myotubular myopathy and a 46,XY disorder of sex development. Patients present with severe form of congenital myopathy and abnormal male genitalia.