DISEASE:
X-linked myotubular myopathy-abnormal genitalia syndrome

NAME:	X-linked myotubular myopathy-abnormal genitalia syndrome
DESCRIPTION:	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.
ORPHACODE:	456328
SYNONYMS:	Xq28 contiguous gene deletion syndrome
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	MAMLD1 MTM1
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

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