DISEASE:
Megaconial congenital muscular dystrophy

NAME:	Megaconial congenital muscular dystrophy
DESCRIPTION:	A rare, genetic, skeletal muscle disease characterized by an early-onset hypotonia, muscle weakness, global developmental delay with intellectual disability, and cardiomyopathy. Congenital structural heart defects and ichthyosiform cutaneous lesions have also been associated. Muscle biopsy shows characteristic enlarged mitochondria located at the periphery of muscle fibers.
ORPHACODE:	280671
SYNONYMS:	Congenital megaconial myopathy Congenital muscular dystrophy due to phosphatidylcholine biosynthesis defect Congenital muscular dystrophy with mitochondrial structural abnormalities
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	CHKB
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

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