DISEASE:
Congenital muscular dystrophy with intellectual disability and severe epilepsy

NAME:	Congenital muscular dystrophy with intellectual disability and severe epilepsy
DESCRIPTION:	A rare, fatal, inborn error of metabolism disorder characterized by respiratory distress and severe hypotonia at birth, severe global developmental delay, early-onset intractable seizures, myopathic facies with craniofacial dysmorphism (trigonocephaly/progressive microcephaly, low anterior hairline, arched eyebrows, hypotelorism, strabismus, small nose, prominent philtrum, thin upper lip, high-arched palate, micrognathia, malocclusion), severe, congenital flexion joint contractures and elevated serum creatine kinase levels. Scoliosis, optic atrophy, mild hepatomegaly, and hypoplastic genitalia may also be associated.
ORPHACODE:	329178
SYNONYMS:	CDG syndrome type Iu CDG-Iu CDG1U CMD with intellectual disability and severe epilepsy Carbohydrate deficient glycoprotein syndrome type Iu Congenital disorder of glycosylation type 1u Congenital disorder of glycosylation type Iu DPM2-CDG

XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	DPM2
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RELATED CONTENT

Related Genetic Tests

- Congenital disorders of glycosylation (79 genes)
- Neuromuscular disorders : congenital & distal myopathy, congenital muscle dystrophy / Limb-girdle muscular dystrophy / Rhabdomyolysis / Myopathy (with prominent contractures) / distal artrogryposis (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- dolichyl-phosphate mannosyltransferase subunit 2, regulatory

Related Gene Panels

- Congenital disorders of glycosylation (79 genes) - KUL
- Neuromuscular disorders (166 genes) - VUB