DISEASE:
Laminin subunit alpha 2-related congenital muscular dystrophy

NAME:	Laminin subunit alpha 2-related congenital muscular dystrophy
DESCRIPTION:	Congenital muscular dystrophy type 1A (MCD1A) belongs to a group of neuromuscular disorders with onset at birth or infancy characterized by hypotonia, muscle weakness and muscle wasting.
ORPHACODE:	258
SYNONYMS:	CMD1A Congenital muscular dystrophy due to laminin alpha2 deficiency Congenital muscular dystrophy type 1A MDC1A Merosin-negative congenital muscular dystrophy
XREF(S):	Orphanet OMIM OMIM ICD-10
ANALYTE(S):	LAMA2
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/193

RELATED CONTENT

Related Genetic Tests

- 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

Related Analytes

- laminin subunit alpha 2

Related Gene Panels

- Neuromuscular disorders (166 genes) - VUB