Belgian Genetic Tests database

DISEASE:
Fatal infantile hypertonic myofibrillar myopathy

NAME:	Fatal infantile hypertonic myofibrillar myopathy
DESCRIPTION:	Fatal infantile hypertonic myofibrillar myopathy is a rare, genetic skeletal muscle disease characterized by muscle stiffness and rigidity, hypertonia, weakness, respiratory distress and normal cognition. Patients have persistently elevated creatine kinase and histopathology is typical of myofibrillar myopathy. The manifestation onset follows the short period of normal infantile development and leads to progressive respiratory insufficiency and early death.
ORPHACODE:	280553
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	CRYAB
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RELATED CONTENT

Related Genetic Tests

- Cardiomyopathy, hereditary (gene panel)
- 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

- crystallin alpha B

Related Gene Panels

- Cardiomyopathy, hereditary (208 genes) - VUB
- Neuromuscular disorders (166 genes) - VUB