# DISEASE:
# Congenital myopathy with excess of thin filaments

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| **NAME:** | Congenital myopathy with excess of thin filaments |
| **DESCRIPTION:** | A rare, genetic, congenital myopathy disorder characterized by variable degrees of muscular weakness, frequently associated with severe nemaline myopathy-like disease (including neonatal hypotonia, lack of spontaneous movements, feeding and swallowing difficulties, frequent respiratory infections, respiratory insufficiency, early death), and histopathologic findings of large, densely packed, subsarcolemmal accumulations of thin, actin-immunopositive filaments (with or without intranuclear nemaline rods) on muscle biopsy. |
| **ORPHACODE:** | 98904 |
| **SYNONYMS:** | Actin myopathy |
| **XREF(S):** | Orphanet<br>OMIM<br>ICD-10 |
| **ANALYTE(S):** | ACTA1 |
| **CREATED:** | 13 May 2019 - 01:02 |
| **CHANGED:** | 22 Jun 2023 - 16:14 |

# 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

- actin alpha 1, skeletal muscle

## Related Gene Panels

- Neuromuscular disorders (166 genes) - VUB