# DISEASE:
# Early-onset myopathy-areflexia-respiratory distress-dysphagia syndrome

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| **NAME:** | Early-onset myopathy-areflexia-respiratory distress-dysphagia syndrome |
| **DESCRIPTION:** | A rare congenital myopathy characterized by early onset of severe muscular weakness, respiratory distress due to diaphragmatic paralysis, dysphagia and areflexia, joint contractures, and scoliosis. Decreased fetal movements are seen in some individuals. Muscle biopsy may show a combination of dystrophic and myopathic features. The clinical course is variable, with some patients becoming ventilator-dependent and never achieving ambulation. |
| **ORPHACODE:** | 439212 |
| **SYNONYMS:** | EMARDD |
| **XREF(S):** | Orphanet<br>ICD-10<br>OMIM |
| **ANALYTE(S):** | MEGF10 |
| **CREATED:** | 13 May 2019 - 01:02 |
| **CHANGED:** | 22 Jun 2023 - 16:14 |

# RELATED CONTENT

## Related Genetic Tests

- Child Interstitial Lung Disease (child - 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
- Centrum Menselijke Erfelijkheid - KUL

## Related Analytes

- multiple EGF like domains 10

## Related Gene Panels

- Neuromuscular disorders (166 genes) - VUB
- chILD (34 genes) - KUL