# DISEASE:
# Neonatal Marfan syndrome

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| **NAME:** | Neonatal Marfan syndrome |
| **DESCRIPTION:** | Neonatal Marfan syndrome is a rare, severe and life-threatening genetic disease, occuring during the neonatal period, characterized by classical Marfan syndrome manifestations in addition to facial dysmorphism (megalocornea, iridodonesis, ectopia lentis, crumpled ears, loose redundant skin giving a 'senile' facial appearance), flexion joint contractures, pulmonary emphysema, and a severe, rapidly progressive cardiovascular disease (including ascending aortic dilatation and severe mitral and/or tricuspid valve insufficiency). Additionally, skeletal manifestations (arachnodactyly, dolichostenomelia, pectus deformities) are also associated. |
| **ORPHACODE:** | 284979 |
| **SYNONYMS:** | Neonatal MFS |
| **XREF(S):** | Orphanet<br>ICD-10 |
| **ANALYTE(S):** | FBN1 |
| **CREATED:** | 13 May 2019 - 01:02 |
| **CHANGED:** | 22 Jun 2023 - 16:14 |

# RELATED CONTENT

## Related Genetic Tests

- Marfan Syndrome

## Related Laboratories

- Centrum Medische Genetica - UZ Gent

## Related Analytes

- fibrillin 1

## Related Gene Panels

- Familiale thoracale aorta aneurysmata (19 genes) - UGent