

Zespół Marfana noworodków

Kod Orpha: 284979 Kod OMIM:

Opis choroby *

Definicja

Neonatal Marfan syndrome is a rare, severe and life-threatening genetic disease, occurring 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.

Dane

Klasyfikacja

Choroba

Synonimy

Neonatal MFS

Noworodkowy MFS

Kod ORPHA

284979

Kod OMIM

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Kod ICD10

Q87.4

Kod ICD11

LD28.0Y

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.