Opis choroby *

Definicja

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.

Dane

Klasyfikacja
Choroba

Synonimy
Neonatal MFS
Noworodkowy MFS

Kod ORPHA
284979

Kod OMIM
-

Kod ICD10
Q87.4

Kod ICD11
LD28.0Y

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*Źródło

orphanet