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 O87.4

Kod ICD11 LD28.0Y

*Źródło

orphanet