Neonatal Marfan syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Neonatal Marfan syndrome. ORPHA:284979

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.