Lethal arteriopathy syndrome due to fibulin-4 deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Lethal arteriopathy syndrome due to fibulin-4 deficiency. ORPHA:314718

Lethal arteriopathy syndrome due to fibulin-4 deficiency is a rare, genetic, vascular disorder characterized by severe aneurysmal dilatation, elongation, and tortuosity of the thoracic aorta, its branches and pulmonary arteries with stenosis at various typical locations, typically resulting in infantile demise. Variable associated features may include cutis laxa, long philtrum with thin vermillion border, hypertelorism, sagging cheeks, arachnodactyly, joint laxity and pectus deformities.