

Zespół tętniaka i zapalenia kości i stawów

Kod Orpha: 284984 Kod OMIM: 613795

Opis choroby *

Definicja

A rare, genetic, systemic disease characterized by the presence of arterial aneurysms, tortuosity and dissection throughout the arterial tree, associated with early-onset osteoarthritis (predominantly affecting the spine, hands and/or wrists, and knees) and mild craniofacial dysmorphism (incl. long face, high forehead, flat supraorbital ridges, hypertelorism, malar hypoplasia and, a raphe, broad or bifid uvula), as well as mild skeletal and cutaneous anomalies. Joint abnormalities, such as osteochondritis dissecans and intervertebral disc degeneration, are frequently associated. Additional cardiovascular anomalies may include mitral valve defects, congenital heart malformations, ventricular hypertrophy and atrial fibrillation.

Dane

Klasyfikacja

Choroba

Synonimy

AOS

Kod ORPHA

284984

Kod OMIM

613795

Kod ICD10

Q87.8

Kod ICD11

LD28.0Y

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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