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