Opis choroby *

Definicja

A rare genetic cardiac malformation characterized by progressive myxomatous degeneration predominantly of the mitral valve (but not uncommonly with multivalvular involvement), presenting as valve thickening and dysfunction with variable stenosis, prolapse, and/or regurgitation, and potentially resulting in lethal heart failure. Hyperextensible skin and joint hypermobility have been reported in some patients. Hemizygous males display a more severe phenotype than heterozygous females.

Dane

Klasyfikacja
Wada morfologiczna

Synonimy
FLNA-related valvular dystrophy
Filamin A-related X-linked myxomatous valvular dysplasia
FLNA-related valvular dystrophy
Filamin A-related X-linked myxomatous valvular dysplasia

Kod ORPHA
555877

Kod OMIM
314400

Kod ICD10
Q23.8

Kod ICD11
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*Źródło

orphanet