

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