Opis choroby *

Definicja

A rare syndromic intestinal malformation characterized by the association of marfanoid features (including marfanoid habitus, severe myopia, retinal detachment, and mitral valve prolapse) with visceral diverticula (inguinal and/or femoral hernia and diverticula of the large and small bowel or urinary bladder). Some patients also had diaphragmatic eventration. There have been no further descriptions in the literature since 1996.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
2464

Kod OMIM
223330

Kod ICD10
Q87.4

Kod ICD11
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*Źródło

orphanet