Opis choroby *

Definicja

MYH9-related disease (MYH9-RD) is an inherited giant platelet disorder with a complex phenotype characterized by congenital thrombocytopenia and possible subsequent manifestations of sensorineural hearing loss, presenile cataracts, elevation of liver enzymes, and/or progressive nephropathy often leading to end-stage renal disease (ESRD). Epstein syndrome, Fechtner syndrome, May-Hegglin anomaly and Sebastian syndrome, previously described as distinct disorders, represent some of the different clinical presentations of MYH9-RD.

Dane

Klasyfikacja
Choroba

Synonimy
MYH9-RD
MYH9-RD
Syndromiczna trombocytopenia związana z MYH9
Zaburzenie związane z MYH9
Zespół związany z MYH9
MYH9-related disorder
MYH9-related syndrome
MYH9-related syndromic thrombocytopenia

Kod ORPHA
182050

Kod OMIM
155100

Kod ICD10
D69.4

Kod ICD11
3B64.01

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*Źródło

orphanet