DISEASE:
MYH9-related disease

NAME:	MYH9-related disease
DESCRIPTION:	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.
ORPHACODE:	182050
SYNONYMS:	MYH9-RD MYH9-related disorder MYH9-related syndrome MYH9-related syndromic thrombocytopenia
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	MYH9
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RELATED CONTENT

Related Genetic Tests

- Nephrotic syndrome, Focal Segmental Glomerulosclerosis (FSGS) , Alport syndrome and podocytopathy (gene panel)
- Trombosis - Hemostasis (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- myosin heavy chain 9

Related Gene Panels

- Nephrotic syndrome, FSGS, Alport syndrome (76 genes) - IPG
- Trombosis - Hemostasis (107 genes) - KUL

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