MYH9-related disease

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. MYH9-related disease. ORPHA:182050

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.