Macrothrombocytopenia and Granulocyte Inclusions with or without Nephritis or Sensorineural Hearing Loss

National Cancer Institute

Source

National Cancer Institute. Macrothrombocytopenia and Granulocyte Inclusions with or without Nephritis or Sensorineural Hearing Loss. NCI Thesaurus. Code C158788.

An autosomal dominant disorder caused by mutation(s) of the MYH9 gene, encoding myosin-9. Clinical features include thrombocytopenia, giant platelets, and characteristic inclusions in peripheral blood leukocytes, and may be associated with other organ dysfunction. It comprises the Epstein syndrome, Fechtner syndrome, May-Hegglin anomaly, and Sebastian syndrome-- all of which were previously believed to be distinct entities.