The Platelet Charity
Helping people with bleeding disorders

MYH9

What is it?

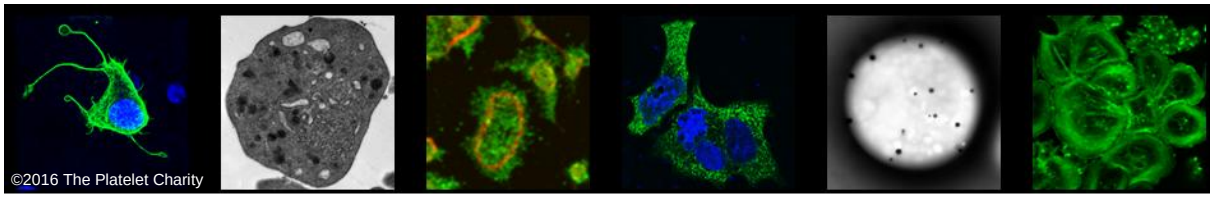
- MYH9 is a mild platelet bleeding disorder which can be diagnosed from childhood to old age.
- It was first described by a German physician Richard May in 1909 and subsequently by a Swiss physician Robert Hegglin in 1945.
- MYH9 was originally described as four distinct clinical conditions: Epstein syndrome, May-Hegglin anomaly, Sebastian syndrome and Fechtner syndrome. Since the discovery that each of these conditions has the same genetic basis, they are now referred to as one condition: ***MYH9-related disorder***. Some of the individual syndromes have other features such as cataracts, hearing problems and kidney problems.
- MYH9 encodes for a protein, myosin-9, which is key component of a protein complex known as myosin IIA. Mutations in the *MYH9* gene cause a reduction in platelet formation.
- Mutations in *MYH9* cause a mild reduction in the number of platelets (thrombocytopenia).

Who suffers?

- MYH9 affects males and females of all age in approximately equal numbers. A single copy of a mutant gene is inherited from the mother or from the father (autosomal dominant).

What are the symptoms?

- Patients with MYH9 disorder may experience nose bleeding, easy bruising, bleeding from gums, heavy or prolonged menstrual bleeding (menorrhagia), bleeding after childbirth, abnormal bleeding after surgery or dental work, and gastrointestinal bleeding.
- Patients with *MYH9* have additional defects which vary between patients:
  - hearing loss (from birth to late adulthood);
  - kidney disease which can lead to end-stage kidney failure (from early adulthood);
  - cataracts (from early adulthood: less common).



Diagnosis

- Patients have a low platelet count (thrombocytopenia) and enlarged platelets; the presence of small rods known as Dohle-like bodies are sometimes seen in white blood cells examined under the microscope.
- Patients may also have hearing loss, kidney disease or cataracts.

Treatment

- Treatment should be led by and discussed with a haematologist with experience in bleeding disorders.
- Treatment is the same as other conditions where there are platelet function defects. General strategies include avoidance of medications that inhibit platelet function further (e.g. aspirin) and compression at sites of injury. Patients may be treated with antifibrinolytics (e.g. tranexamic acid) or platelet transfusion prior to surgery.
- Patients should carry a bleeding disorders / haemorrhagic states card (issued by a Haemophilia Care Centre) to alert others to their condition.
- Genetic counselling may be offered.

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