The Platelet Society

MYH9 related disorder

What is it?

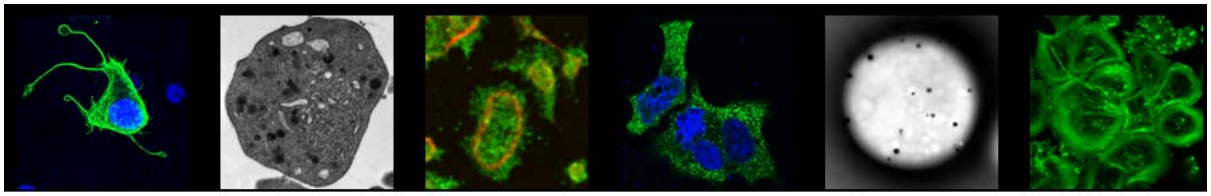
- **MYH9** is a mild platelet bleeding disorder which can be diagnosed from childhood to old age.
- It was first described by a German physician Richard May in 1909 and subsequently by a Swiss physician Robert Hegglin in 1945.
- **MYH9** was originally described as four distinct clinical conditions: 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, a key component of a protein complex called myosin IIA, which is important in cell movement and maintaining cell shape.
- Mutations in the *MYH9* gene cause a mild reduction in the number of platelets (thrombocytopenia) and increased platelet size.

Who suffers?

- MYH9 affects males and females of all age in approximately equal numbers.
- The condition is inherited in an autosomal dominant pattern, which means that just one copy of the altered gene, inherited from the mother or father is sufficient to cause the disorder.
- Approximately 30% of cases result from new mutations, in patients with no family history of the disorder

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).



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Diagnosis

- Patients have a low platelet count (thrombocytopenia) and enlarged platelets; clumps of non-functional Myosin 9 protein (called inclusion bodies) are present in white blood cells and can be observed using a 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, according to the severity of bleeding manifestations.
- 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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