The Platelet Charity
Helping people with bleeding disorders

GFI1B

What is it?

- GFI1B-related thrombocytopenia is a rare, platelet bleeding disorder which can be diagnosed from childhood to old age.
- It is caused by a mutation in the gene for the Growth Factor Independent 1B (*GFI1B*) transcription factor.
- The first family with a platelet disorder caused by a mutation in *GFI1B* was identified by Drs Marie-Christine Morel-Kopp, William Stevenson and Chris Ward in Melbourne in 2013.
- Less than ten patients have been identified worldwide with a mutation in *GFI1B*.
- GFI1B is a protein that controls the expression of a number of other proteins involved in the production of platelets from the platelet mother cell, the megakaryocyte.
- Mutations in *GFI1B* causes a mild to moderate reduction in the number of platelets (thrombocytopenia) and a variable defect in platelet function.

Who suffers?

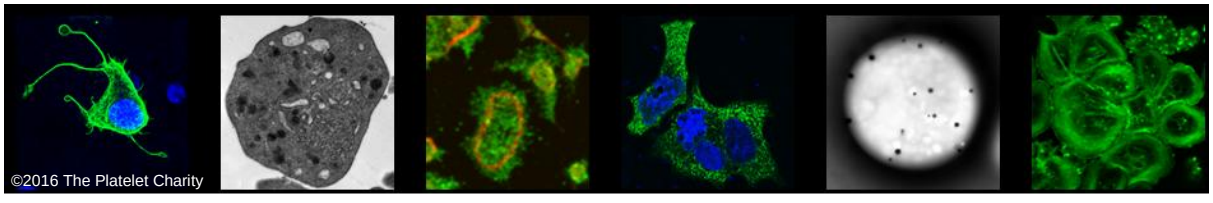
- Males and females are equally affected. A single copy of the mutated gene can be inherited from the mother or father. It can be identified from early childhood into old age.

What are the symptoms?

- Patients with a *GFI1B* mutation may experience nose bleeds, easy bruising, bleeding from gums, heavy or prolonged menstrual bleeding (menorrhagia), bleeding after childbirth, abnormal bleeding after surgery or dental work, and gastrointestinal bleeding.

Diagnosis

- Patients have a mild reduction in platelet count (thrombocytopenia) and reduced platelet aggregation (clumping) in response to several platelet stimuli.
- Some patients have large platelets and reduced α-granules in their platelets which causes them to appear grey on a blood film (n.b. this disorder is distinct from Grey platelet syndrome).
- A definitive diagnosis of GFI1B is made by genetic sequencing.



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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