The Platelet Society

FLI1 and Paris-Trousseau syndrome

What is it?

- **FLI1** (pronounced “Fly-one") is a rare, platelet bleeding disorder which can be diagnosed at any age.
- It is caused by a mutation a gene called ‘Friend leukaemia integration 1 transcription factor’ (*FLI1*).
- The gene is also completely absent (along with several other genes) in patients with **Paris-Trousseau syndrome** which is caused by partial loss of chromosome 11.
- Both conditions are extremely rare with less than 100 hundred patients identified worldwide.
- FLI1 is a protein that controls the production of a number of other proteins involved in the production of platelets from the platelet mother cell, the megakaryocyte.
- A mutation in FLI1 causes a mild to moderate reduction in the number of platelets present in the blood (thrombocytopenia) and a variable defect in platelet function.

Who suffers?

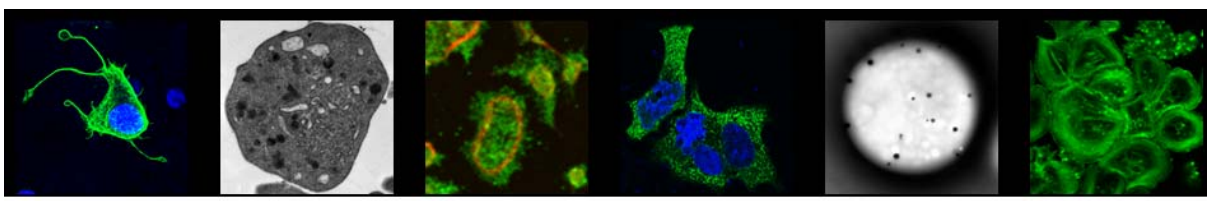
- Males and females are equally affected. In most cases, a single copy of the mutated gene can be inherited from the mother or father. It can be identified from early childhood into old age.

What are the symptoms?

- Patients with a FLI1 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 to moderate reduction in platelet count (thrombocytopenia), large platelets, abnormally large storage granules within the platelets (which are few in number) and reduced platelet aggregation (clumping) in response the molecules that trigger blood clotting.
- Diagnosis 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 anti-fibrinolytics (e.g. tranexamic acid) or platelet transfusion prior to surgery.
- Patients should carry a bleeding disorders / haemorrhagic status card (issued by a Haemophilia Care Centre) to alert others to their condition.
- Genetic counselling may be offered.

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