The Platelet Society

RUNX1 (familial platelet disorder)

What is it?

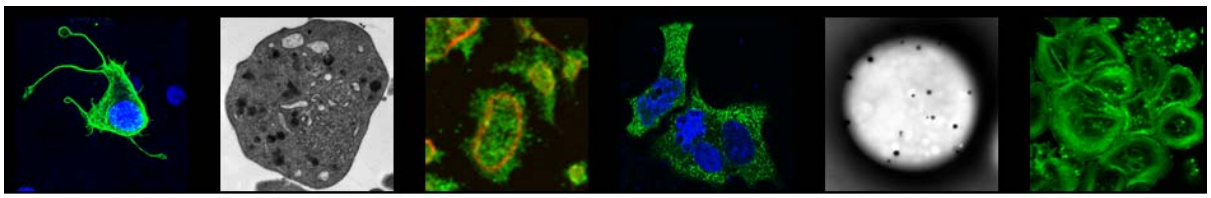
- **RUNX1** (also known as **familial platelet disorder with predisposition to acute myeloid leukemia, FPD/AML**) is a rare, mild bleeding disorder which can be diagnosed at any age.
- **RUNX1** is caused by a mutation in the Runt-related transcription factor 1 (*RUNX1*) gene.
- The name, **FPD** was first used in 1985 by Frederick Li and colleagues to describe a large family with a platelet disorder. The link between **FPD**, **AML** and mutations in *RUNX1* gene was made later in 1999.
- **RUNX1** is a rare disorder with over one hundred patients identified worldwide.
- *RUNX1* is a transcription factor responsible for controlling the expression of proteins involved in the production of various blood cell types including platelets.
- Mutations in *RUNX1* causes a mild reduction in the number of platelets (thrombocytopenia) and a mild 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 *RUNX1* 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.
- There is a link between being diagnosed with a **RUNX1 bleeding disorder** and being at risk of developing leukaemia or changes in the bone marrow (myelodysplastic syndrome); however most patients with this platelet defect do not develop leukaemia. Your consultant will discuss this with you.



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Diagnosis

- Patients have a mild to moderate reduction in platelet count (thrombocytopenia) and reduced platelet aggregation (clumping) in response to several platelet stimuli.
- Platelets from patients with a **RUNX1 disorder** express a protein that is normally absent in platelets, MYH10: this can only be investigated in a specialist testing laboratory.
- A definitive diagnosis of **RUNX1** 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, 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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