The Platelet Charity
Helping people with bleeding disorders

RUNX1 (familial platelet disorder)

What is it?

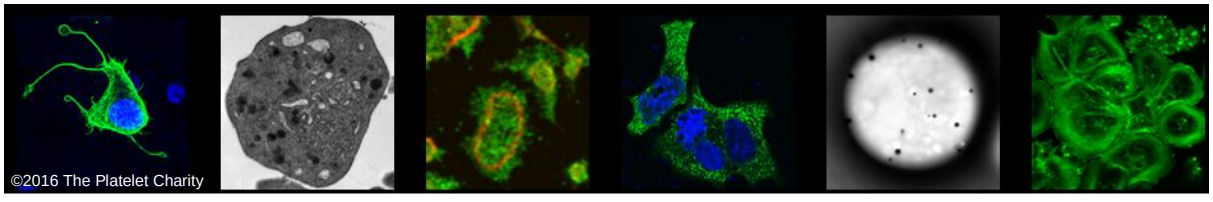
- RUNX1 (also known as *familial platelet disorder with predisposition to acute myeloid leukemia*, FPD/AML) is a rare, mild bleeding disorder which can be diagnosed from childhood to old age.
- RUNX1 is caused by a mutation in the Runt-related transcription factor 1 (RUNX1) gene.
- The name, FPD, was first used in 1985 by Frederick Li and colleagues in Harvard to describe a large family with a platelet disorder. The link between FPD, AML and mutations in RUNX1 gene was made in 1999.
- RUNX1 is a rare disorder with over one hundred patients identified worldwide.
- RUNX1 is a protein that controls the expression of a number of proteins that are 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.



Diagnosis

- Patients have a mild 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. 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.

The Platelet Charity 2017