The Platelet Charity
Helping people with bleeding disorders

Schlafen14

What is it?

- Schlafen14 (SLFN14) disorder is a recently discovered rare mild bleeding disorder that is characterised by a low platelet count (thrombocytopenia). It can be diagnosed from childhood to old age.
- It is caused by mutations in the gene *SLFN14* which encodes for a protein that regulates expression of other proteins in the platelet mother cell, the megakaryocyte.
- It was identified in 2015 in the University of Birmingham (UK) by the geneticist, Neil Morgan.
- There are less than ten affected families worldwide with the majority in the UK.
- A mutation in *SLFN14* causes a mild reduction in the number of platelets (thrombocytopenia) and a mild defect in platelet function.
- SLFN14 is only found in platelets and the platelet mother cell, the megakaryocyte.

Who suffers?

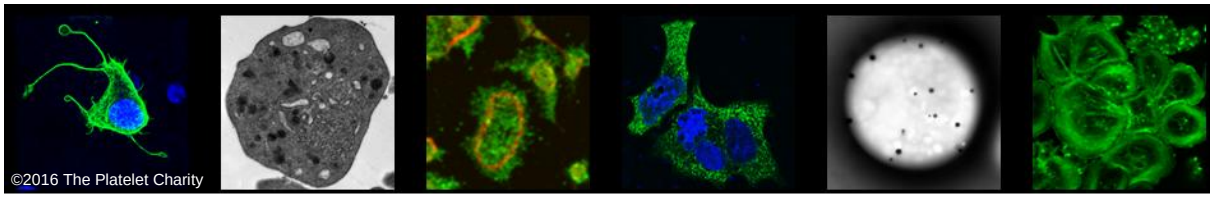
- SLFN14 disorder affects males and females of all ages in approximately equal numbers. A single copy of a mutant gene is inherited from the mother or father (autosomal dominant).

What are the symptoms?

- Patients with SLFN14 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.

Diagnosis

- Patients have a mild reduction in platelet count (thrombocytopenia), slightly enlarged platelets and reduced platelet aggregation (clumping) in response to several platelet stimuli.
- Platelets have a reduced number of a particular type of storage granule in their cytoplasm known as dense granules, which release stimuli that reinforce platelet activation.
- The reduction in the number of dense granules can be shown using electron microscopy which is only available in specialised research laboratories.
- A definitive diagnosis of *SLFN14* 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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