The Platelet Society

Glanzmann thrombasthaenia

What is it?

- **Glanzmann thrombasthaenia** (**GT**) is a well characterised and researched severe platelet bleeding disorder.
- The first patient was identified in 1918 by the German physician Eduard Glanzmann.
- **GT** is caused by a loss of a complex of two proteins on the platelet surface. The proteins are GPIIb and GPIIIa, and together they form the GPIIb-IIIa complex (which is also known as integrin αIIbβ3).
- The GPIIb-GPIIIa complex is only found in platelets and the platelet mother cell, the megakaryocyte.
- GPIIb-IIIa binds to several proteins in the blood. The best know is fibrinogen, which helps platelets to clump together to form blood clots. When GPIIb-IIIa is missing, the platelet cannot bind fibrinogen, so the platelets cannot clump together.

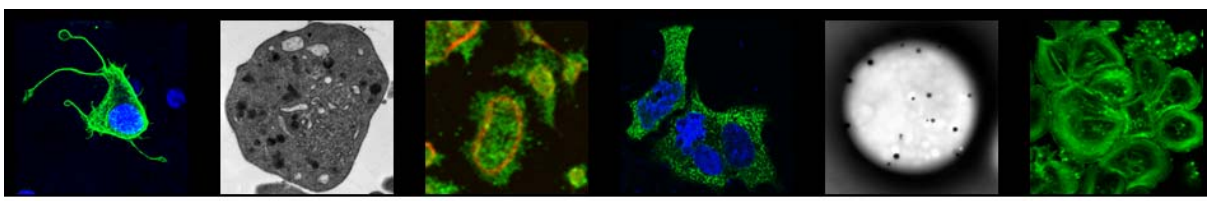
Who suffers?

- **GT** is usually detected at an early age, especially where other family members are known to be affected.
- **GT** affects males and females of all age in approximately equal numbers. Two copies of the mutant gene are inherited (one each) from the mother and father (autosomal recessive).
- It is more common in families were there are relationships between individuals who are blood relatives of each other. In general, carriers of the condition (who only have one mutated gene) do not experience any clinical problems.
- There are over one thousand patients worldwide, with over fifty patients in the UK.

What are the symptoms?

- Patients with **GT** 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 normal platelet count and normal platelet size, but they are unable to undergo platelet aggregation (clumping) upon activation by all stimuli that are used to investigate platelet function, with the exception of ristocetin.
- Diagnosis is made (by a technique called flow cytometry) using an antibody against normal GPIIb (also known as CD41).
- A definitive 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 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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