The Platelet Society

Adenosine diphosphate (ADP) $P2Y_{12}$ receptor

What is it?

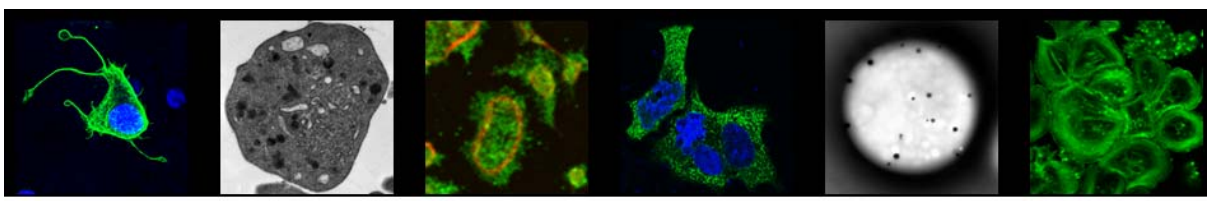
- Adenosine diphosphate (ADP) is a chemical that is released by platelets to help them form blood clots. It binds to the surface of platelets through two different protein receptors called $P2Y_1$ and $P2Y_{12}$. If these receptors are not working correctly it can lead to excessive bleeding.
- **$P2Y_{12}$ disorder** is a rare mild platelet bleeding disorder which can be diagnosed at any age.
- The first patient with a $P2Y_{12}$ disorder was described by Dr Marco Cattaneo in Italy in 1992.
- There are less than 50 patients worldwide, with approximately 10 patients in the UK.
- **$P2Y_{12}$ disorder** is caused by mutations in the *P2Y12* gene which results in a loss or a dysfunctional $P2Y_{12}$ receptor.
- The $P2Y_{12}$ receptor is found in platelets and the platelet mother cell, the megakaryocyte. It is also found in a small number of other tissues where its role is unknown.

Who suffers?

- **$P2Y_{12}$ disorder** affects males and females of all ages in 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 clinical problems, although there are exceptions.

What are the symptoms?

- Patients with a **$P2Y_{12}$ 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.



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Diagnosis

- Patients have a normal platelet count and normal platelet size.
- They have a reduced aggregation (clumping) response to ADP, one of the stimuli used to test platelet function.
- 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.

The Platelet Society 2018