The Platelet Society

Wiskott-Aldrich syndrome and X-linked thrombocytopenia

What is it?

- **Wiskott-Aldrich syndrome** (**WAS**) is a rare immune system disorder which is associated with a mild bleeding disorder. It is usually diagnosed in the first year of life due to the increase in susceptibility to infection.
- **WAS** was first identified by Dr Alfred Wiskott in 1937.
- **WAS** is caused by a mutation in the *WAS* gene which encodes for WAS protein (WASp). WASp is important for the structure of blood cells and for allowing them to change shape and move.
- Some mutations in the *WAS* gene give rise to a milder form of the disorder, known as **X-linked thrombocytopenia** (**XLT**) which is characterized primarily by a mild bleeding disorder.

Who suffers?

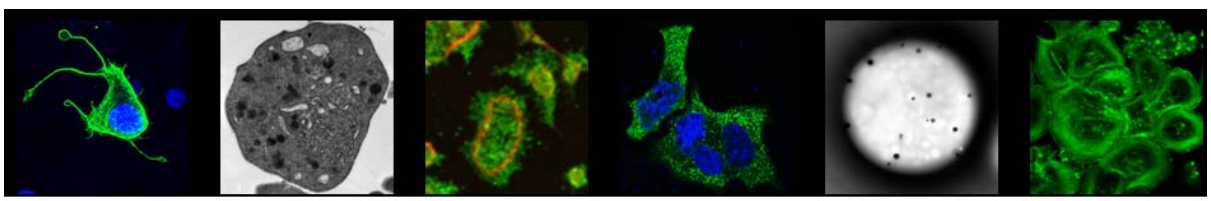
- **WAS** occurs in approximately 1 in 250,000 males; it is extremely rare in females. The mutant gene is inherited on the X sex chromosome from the mother or father. Men have one X chromosome and women have two. Females with one copy of a mutant *WAS* gene are usually symptomless due to compensation of the other *WAS* gene. These females are referred to as 'carriers'.

What are the symptoms?

- Patients with **WAS** 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.
- Patients are susceptible to infections which can be life-threatening.

Diagnosis

- Patients have a reduced platelet count (thrombocytopenia) and platelet size. Platelet activation is normal but platelet clumps fall apart in flowing blood.
- Patients have a reduced level of circulating antibodies and reduced white cell function.
- Diagnosis is based on genetic sequencing of the *WAS* gene.



The Platelet Society

Treatment

- For treatment and prevention of infections, treatment should be led by and discussed with a consultant in immunology.
- For bleeding, 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 anti-fibrinolytics (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.
- Bone marrow transplantation can be used to cure **WAS**. This is a major procedure that requires counselling.

The Platelet Society 2018