The Platelet Charity
Helping people with bleeding disorders

Hermansky-Pudlak syndrome (HPS)

What is it?

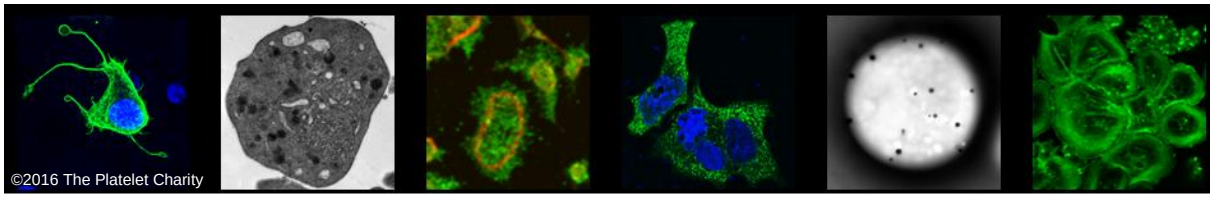
- HPS is a group of rare mild platelet bleeding disorders which can be diagnosed from childhood to old age.
- The first patient was described by two Czechoslovakian pathologists, Frantisek Hermansky and Paulus Pudlak, in 1959.
- HPS is known to be caused by mutations in 9 genes which encode for proteins that are key components of storage granules in platelets and several other cell types. In platelets, the granules are called dense granules.
- Upon platelet activation, dense granules release chemicals such as ADP which reinforce activation. Granules in other cells control a number of factors including hair and eye colour.
- HPS-1 is the most frequent of the HPS disorders with several hundred patients worldwide. HPS-1 is the most frequent genetic disorder in Puerto Rico. Several HPS mutations have been found in just one or two families (e.g. HPS-7, HPS-8 and HPS-9).

Who suffers?

- HPS 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 with blood relatives. In general carriers of the condition (who only have one mutated gene) do not experience clinical problems.

What are the symptoms?

- Patients with HPS 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.
- Some but not all forms of HPS can lead to inflammation of the bowel (called granulomatous colitis) or scarring of the lungs (called pulmonary fibrosis). These typically present in patients between the ages of 40-50 and cause premature death. Patients with other forms of HPS have a normal life expectancy.



Diagnosis

- Patients have a normal platelet count and normal platelet size but are unable to release the contents of dense granules on activation. This can be shown by measurement of ATP or serotonin secretion: these tests are only performed in a handful of clinical test centres.
- Patients with HPS have light hair and eye colour (hypopigmentation).
- Diagnosis is made by the absence of dense granules on electron micrographs.
- A definitive diagnosis of HPS 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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