



My46 Trait Profile

Hermansky-Pudlak syndrome

Hermansky-Pudlak syndrome is a rare genetic condition characterized by albinism of the eyes and skin, prolonged or easy bleeding, and in some types, progressive lung disease. It is caused by mutations in one of many different genes.

Characteristics of Hermansky-Pudlak syndrome

Hermansky-Pudlak syndrome (HPS) includes the presence of three main features: albinism affecting the eyes and skin; tendency to bleed easily; lung disease. In HPS, individuals have albinism that affects both eyes and skin, known as oculocutaneous albinism. People with HPS may have very little skin, eye, or hair coloring, or they may have olive skin, brown hair, and brown eyes just a shade lighter than the rest of their family. Eye problems include low vision and eye movement problems (i.e., horizontal nystagmus, where the eyes seem to dart side-to-side uncontrollably). Although most people with HPS have some vision, they are considered legally blind.

People with HPS also have a bleeding tendency such that they can bleed easily from minimal accidents, and bleeding can be difficult to stop. Normally, platelets in the blood clump together to stop bleeding, such as from a cut or when a child's tooth falls out. People with HPS have a typical number of blood platelets, but the platelets are unable to bond together well. People with HPS also bruise easily.

Individuals with HPS may also have storage problems within the cells that can lead to lung fibrosis or a type of colitis that can be hard to treat. People with certain forms of HPS (i.e., HPS1 or HPS4) often develop progressive pulmonary fibrosis (PF), usually in their 30s or 40s. This is a major cause of early death in HPS, within about ten years from the start of fibrosis. Those with another form of HPS (i.e., HPS2) are prone to infections. Intellectual development is not affected by HPS.

Diagnosis/Testing

HPS should be suspected in anyone with albinism and bleeding diathesis. Albinism may be subtle, and can include individuals with olive skin and brown eyes. With a light, a doctor can look at pigment in the retina to determine if eyes are affected. Lack of delta bodies in platelets, which can be seen by electron microscopy, is the most accurate test for HPS. Genetic testing is also available, and is the only method to distinguish sub-types. The genes in which changes or mutations are known to cause HPS are HPS1, AP3B1, HPS3, HPS4, HPS5, HPS6, DTNBP1, BLOC1S3, and BLOC1S6. Individuals with HPS with Puerto Rican ancestry tend to have mutations in the HPS1 or HPS3 genes.

Management/Surveillance

As with other types of albinism, skin protection is also important for individuals with HPS. People with HPS need to minimize direct sun, cover the skin with clothing and hats, use high SPF sunscreen, and watch closely for changes that may indicate skin cancer. Dark glasses can protect the eyes, but may also reduce vision further. Assistive devices for low vision, such as magnifiers, large print books, or guide dogs may be helpful.

During procedures such as tooth extractions or surgeries, bleeding tendency can be managed with a medication called DDAVP (also called desmopressin), or in some cases, transfusions. It is recommended that individuals with HPS avoid medications that cause increased bleeding or that limit blood clotting, such as aspirin or NSAIDs. Medical alert bracelet that specifies "platelet defect" can be important for emergency treatment.

Keeping lungs as healthy as possible by avoiding air pollutants, quickly treating lung infections, and exercising

regularly can help delay the effects of lung fibrosis for those with certain forms of HPS (i.e., HPS1 and HPS4). HPS-related inflammatory bowel disease shares some characteristics with Crohn's colitis. Medication and changes in eating habits can sometimes relieve symptoms. Any rectal bleeding requires evaluation.

Mode of inheritance

HPS is inherited in an autosomal recessive pattern. This means that an individual has to inherit two mutations (i.e., one from each parent) to be affected with HPS. If both parents are carriers of HPS, they have a 1 in 4 (25%) chance with each pregnancy of having a child with HPS.

Risk to family members

Parents of a child with HPS are carriers of HPS. If a sibling of a child with HPS is unaffected, he/she has a 2 in 3 (66%) chance of being a carrier of HPS.

Special considerations

None

Resources

Genetics Home Reference: Hermansky-Pudlak syndrome

<http://ghr.nlm.nih.gov/condition/hermansky-pudlak-syndrome>

Hermansky-Pudlak Syndrome Network

<http://www.hpsnetwork.org/>

NOAH: Hermansky Pudlak Syndrome

<http://www.albinism.org/publications/HPS.html>

References

Gahl WA, Huizing M. (Updated 28 February 2013). Hermansky-Pudlak Syndrome. In: GeneReviews at GeneTests Medical Genetics Information Resource (database online). Copyright, University of Washington, Seattle. 1997-2013. Available at <http://www.ncbi.nlm.nih.gov/books/NBK1287/>. [Accessed 10/02/2013].

[Seward, SL. et al. \(2013\).](#) "Hermansky-Pudlak Syndrome: Health Care Throughout Life." *Pediatrics* 132(1):153-160.

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