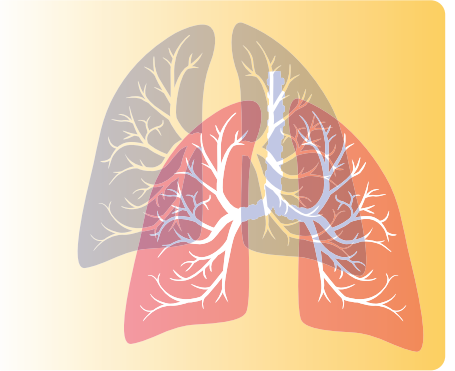


GENETIC DISORDERS SERIES

## What is Hermansky-Pudlak Syndrome?

Hermansky-Pudlak Syndrome (HPS) is a rare inherited disease, named after two doctors in Czechoslovakia who, in 1959, recognized similar health conditions in two unrelated adults. Since the discovery of HPS, the condition has occurred all over the world but is most common in Puerto Rico. The most common health conditions with HPS are albinism, the tendency to bleed easily, and pulmonary fibrosis. A growing number of gene mutations have been identified causing HPS (including numbers HPS1 to HPS10).



### What is albinism?

Albinism is an inherited condition in which reduced pigmentation (coloring) is present in the body. As a result, people with albinism are often fair-skinned with light hair. However, skin, hair, and eye color may vary, as some people with albinism may have dark brown hair and green or hazel/brown eyes. Low vision and various degrees of nystagmus (uncontrolled eye movement from side to side) is seen in all cases. All people who have HPS have albinism, but not all people with albinism have HPS.

### Why do people with HPS bleed easily?

Platelets are special cell fragments that circulate in the blood stream and help the blood to clot. HPS patients have normal numbers of platelets, but they are not made correctly and do not function well, so the blood does not clot properly. As such, persons with HPS may bruise easily or have frequent or heavy nose bleeds. Bleeding problems are usually mild, but in some cases they can be quite serious, especially in certain situations such as surgery. Some women might require special medical attention during their menstrual cycle or at childbirth because they can lose too much blood.

### What causes pulmonary fibrosis in HPS?

The exact cause of pulmonary fibrosis in HPS is still uncertain. In HPS, there is inflammation present, and over time, the lungs become scarred or fibrosed. The fibrosis in the lungs limits the ability for oxygen to enter the blood. This results in a lower than normal amount of oxygen to reach the cells of the body. Damage to other body organs may occur if oxygen levels are severely low or continue to be low and uncorrected over a long time. Besides a low blood oxygen level, symptoms of pulmonary fibrosis can include shortness of breath and fatigue. Pulmonary fibrosis develops during early adulthood and gradually worsens as a person ages.

### What other problems are common in HPS?

**Intestinal problems**—Approximately 15% of individuals with HPS develop significant colitis (inflammation) in the intestines. This condition, similar to Crohn's disease, can

cause abdominal pain and bloody diarrhea. Sometimes these problems may be severe enough to require surgery or a blood transfusion. Intestinal problems usually develop in teens, but they may develop in younger children.

**Kidneys and heart problems**—Less common than lung and intestinal problems are problems in the kidneys and heart. The heart is under stress with pulmonary fibrosis due to abnormal lung function and high blood pressure (pulmonary hypertension). It can also be stressed if there is not enough oxygen in the body.

**Vision problems**—Albinism causes HPS patients to have reduced eye pigment; during early eye development, the eye and surrounding nerves can grow abnormally. This can lead to poor vision, the development of crossed-eyes (strabismus), and nystagmus. It is common with HPS to be sensitive to light (photophobia) since there is little pigment in the eyes to protect against sunlight.

**Skin problems**—The reduction of pigmentation in the skin from albinism results in an increased chance of developing skin problems. Fair skin can be easily damaged by the sun.

### How is HPS diagnosed?

Many physicians are not familiar with HPS and its symptoms because it is such a rare condition. For a child with albinism it is important to ask the child's healthcare provider about the possibility of HPS.

All patients with HPS have albinism. Because of the vision problems associated with albinism, the most common diagnostic test for albinism is an eye examination. The skin and hair may also be examined for low amounts of coloring or pigment. To look for HPS, the person's blood is examined under an electron microscope to look at the platelets. In HPS, the platelets do not appear normal because they lack dense bodies. Not seeing dense bodies in platelets is a characteristic feature of HPS. Blood testing can be done for some HPS genes but a person can have HPS even if one of the known gene mutations is not found.

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## How is Hemansky-Pudlak Syndrome managed and treated?

While there is no cure for HPS, the disease can be managed through proper medical care. Patients can be instructed in self-management strategies to help manage their condition. Because multiple organs of the body can be affected by HPS, patients with HPS should be cared for by a team of healthcare providers. Team members include:

**Pulmonologist** (*lung doctor*)—Since pulmonary fibrosis can be a serious problem, HPS patients should be cared for by a pulmonologist with lung function testing starting by age 12 years or sooner if they experience shortness of breath. They should receive lung function tests (PFTs) each year to monitor lung function. All patients with HPS should get a yearly flu vaccine and a pneumococcal pneumonia vaccine. Lung function gets worse over time and may limit a person's daily activities. Oxygen is used in people with HPS who have low oxygen saturation levels with sleep or all the time. For additional information about PFTs or oxygen therapy, go to [www.thoracic.org/patients/](http://www.thoracic.org/patients/)

**Hematologist** (*blood doctor*)—HPS patients should visit a hematologist and learn how to manage their bleeding risk. As a precaution, aspirin or aspirin-related drugs (NSAIDs—non steroidal anti-inflammatory preparations such as ibuprofen, indomethacin, naproxen, sulindac.) should not be taken as they thin the blood and can increase bleeding. Patients who have frequent nosebleeds may benefit from a humidifier to moisten the air in their home.

**Ophthalmologist** (*eye disease doctor*)—It is important for HPS patients to be examined each year for eye conditions and to have their vision checked because their eyes are more sensitive to light; without proper care they develop further vision problems.

**Dermatologist** (*skin doctor*)—Patients with HPS have an increased risk for developing skin disease and skin cancer. Because of their fair, sensitive skin, they should visit their dermatologist each year.

**Gastroenterologist** (*digestive system doctor*)—HPS patients with intestinal problems should consult a gastroenterologist for treatment. Changes in diet and anti-inflammatory medications, such as steroids, may reduce pain and inflammation.

**Gynecologist** (*female reproductive system doctor*)—Female patients with HPS may have excessive menstrual bleeding. They may choose to take oral contraceptives (birth control) to shorten or reduce the menstrual period. Occasionally, medical procedures may be required to reduce the bleeding.

**Geneticist** (*a doctor who specializes in inherited diseases and problems due to gene mutations*)—People with HPS and their family members may wish to consult a geneticist to determine the probability of having children with HPS or to help their children or relatives decide on the need to be tested.

## How will HPS affect my lifestyle?

Many HPS patients can lead relatively normal lives. The biggest challenges for many individuals who are affected by HPS are poor vision, fair skin, the social stigma that can be associated with albinism. Because individuals with HPS have sensitive eyes and skin, they are very sensitive

to sunlight and should take extra precautions to limit sun exposure and to protect themselves when outdoors by wearing sunscreen, sunglasses, and clothing that covers most of their bodies.

Many people with HPS are legally blind, so they may have to use special aids to help them see well. For instance, normal reading glasses or contact lenses as well as hand-held magnifiers or special small telescopes can be helpful in improving general vision. However, some vision problems cannot be corrected with glasses. Many individuals are unable to drive.

HPS patients may develop problems breathing in their thirties and forties. Because scarring of the lung worsens as a person ages, HPS patients may need to use oxygen and feel increasingly tired or short of breath. Smoking tobacco can make symptoms worse so HPS patients should not smoke and should avoid being around smoke.

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## Rx Taking Action

- ✓ If you have albinism and problems with bleeding and/or breathing, ask your health care provider if you should be tested for HPS
- ✓ If you (or your child) have HPS, be sure you see your health care provider regularly and decide together what other specialists you need to see.
- ✓ Because you may have a serious bleeding problem, avoid activities where there is a high likelihood of injury (skateboarding, motorcycles, etc.). Discuss with your health care provider if there are other sports or activities that may need to be avoided due to risk of bleeding.
- ✓ Some medications such as aspirin or ibuprofen can aggravate bleeding. Ask your health care provider or pharmacist before taking these medications.
- ✓ Get regular lung function testing.
- ✓ Do not smoke and avoid all tobacco smoke exposure.

**Doctor's Office Telephone:**

For additional information:

**Hermansky-Pudlak Syndrome Network, Inc.**

<http://www.hermansky-pudlak.org>

<http://www.hpsnetwork.org>

**The National Organization for Albinism and Hypopigmentation**

<http://www.albinism.org>

**National Association for Visually Handicapped**

<http://www.navh.org>

**American Foundation for the Blind (AFB)**

[www.afb.org](http://www.afb.org)

**National Association for Parents of Children with Visual Impairments**

<http://www.napvi.org>