

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 often seen 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).

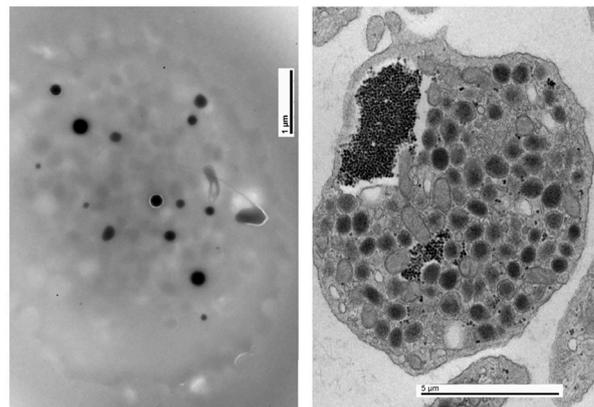


Figure 1. Normal platelet with dense bodies visualized by electron microscopy.

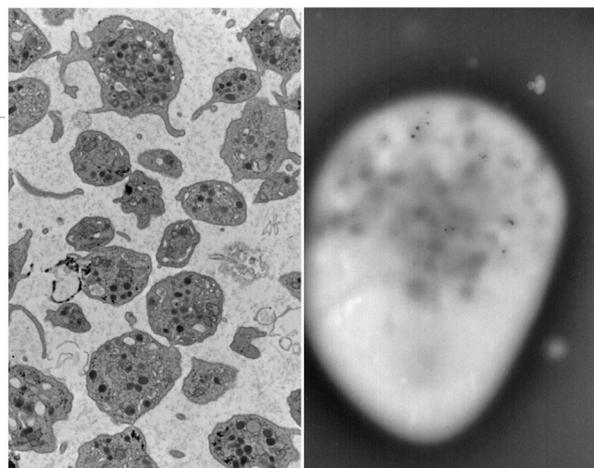


Figure 2. Patient's platelet with virtually absent dense bodies visualized by electron microscopy.

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.

People with albinism all have low vision and varying degrees of nystagmus. All people who have HPS have albinism, but not all people with albinism have HPS.

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

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 and have other issues such as frequent or heavy nose bleeds. Bleeding problems can be mild, but in some cases they can be quite serious, especially in certain situations such

as surgery or injuries. Some women with HPS require special medical attention because they can lose too much blood during their menstrual cycle or at childbirth.

What is pulmonary fibrosis and what causes it in HPS?

Pulmonary Fibrosis is a group of diseases in which there is scarring in the lungs. The exact cause of pulmonary fibrosis in HPS is still uncertain. 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 in HPS occurs in those individuals with HPS₁, HPS₂ and HPS₄. Pulmonary fibrosis in HPS develops in early adulthood and gradually worsens with age.

What are other problems that are seen in HPS?

Intestinal problems—Approximately 15% of people with HPS develop colitis (inflammation) in the intestines. This condition can cause abdominal pain and bloody diarrhea. Sometimes these problems may be severe enough to require surgery or blood transfusions. Intestinal problems usually develop in teens, but they may develop in younger children.

Kidneys and heart problems—Some people with HPS may also develop issues with their heart or kidneys. The heart is under stress with pulmonary fibrosis due to low oxygen levels and can cause pulmonary

hypertension (high blood pressure in the lungs).

There is a subset of HPS individuals that seem to develop a chronic kidney insufficiency. Rarely it has caused the need for both a lung and kidney transplant. At present, the cause is unknown.

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

How is HPS diagnosed?

Many healthcare providers are not familiar with HPS and its symptoms because it is such a rare condition. If a child has albinism it is important to ask the child's healthcare provider to screen for HPS.

Diagnosing HPS begins with establishing the condition of albinism through an eye exam. The skin and hair may be examined for lower amounts of coloring in comparison to the rest of the family. Once albinism is confirmed then HPS can be diagnosed. 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 which is a characteristic feature of HPS. Genetic 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.



How is Hemansky-Pudlak Syndrome managed and treated?

While there is no cure for HPS, the disease can be managed through proper medical care and education to stay as healthy as possible. Because multiple organs of the body can be affected, people 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 people should be cared for by a pulmonologist. Children should be evaluated by a pediatric pulmonologist to establish care, discuss any respiratory concerns with the family, including prevention of pulmonary infections, and consider spirometry if they experience shortness of breath, fatigue or other respiratory complaints. Lung function tests (PFTs) are repeated at least once each year to watch for lung problems. Lung function can get worse over time and may limit a person's daily activities. All people with HPS should get a yearly flu vaccine and a pneumococcal pneumonia vaccine as recommended by one's healthcare provider. Oxygen is used in people with HPS who have low oxygen saturation levels. For additional information about PFTs or oxygen therapy, go to www.thoracic.org/patients/

Hematologist (blood doctor)—People with HPS 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 affect blood clotting and can increase bleeding. People who have frequent nosebleeds may benefit from a

humidifier to moisten the air in their home.

Ophthalmologist (eye disease doctor)—It is important for people with HPS to be examined each year. Their eyes are more sensitive to light and without proper care they develop further vision problems.

Dermatologist (skin doctor)—People 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)—People with HPS with intestinal problems should consult a gastroenterologist for treatment. Changes in diet and anti-inflammatory medications may reduce pain and inflammation.

Gynecologist (female reproductive system doctor)—Women with HPS may have excessive menstrual bleeding. They may choose to take oral contraceptives (birth control) to shorten or reduce the menstrual period. Sometimes, medical procedures may be needed 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 the need for further testing.

How will HPS affect my lifestyle?

Many people with HPS can lead relatively normal lives. A significant challenge for many who are affected by HPS are poor vision, fair skin, and the social stigma that can be associated with albinism. They are very sensitive to sunlight and should take extra precautions to limit sun exposure and to protect themselves when outdoors by wearing appropriate SPF 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 better. For instance, reading glasses, contact lenses, hand-held magnifiers or special small telescopes can be helpful in improving general vision. Their low vision cannot be totally corrected with glasses. Many are unable to drive.

People with HPS may develop problems breathing in their thirties and forties. Because scarring of the lung worsens as a person ages, those with pulmonary fibrosis may feel increasingly tired or short of breath and may need to use oxygen. Antifibrotics and lung transplantation are presently the only treatment options. Smoking or vaping tobacco or marijuana or being exposed to smoke or vape can make symptoms worse so people with HPS should not smoke and should avoid being around smoke.

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R_x Action Steps

- ✓ If you have albinism and problems with bleeding and/or breathing, ask your healthcare provider to test you for HPS.
- ✓ If you (or your child) have HPS, be sure you see your healthcare 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 worsen your risk of bleeding. Ask your healthcare provider or pharmacist before taking these medications.
- ✓ Get regular lung function testing.
- ✓ Do not smoke and avoid all tobacco smoke exposure.

Healthcare Provider's Contact Number:

For More Information

American Thoracic Society

- <https://www.thoracic.org/patients>

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

National Association for Parents of Children with Visual Impairments

- <http://www.napvi.org>

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