



Information about Albinism

National Organization for Albinism and Hypopigmentation

Hermansky-Pudlak Syndrome

Hermansky-Pudlak Syndrome (HPS) is a type of albinism which includes a bleeding tendency and lung disease. HPS may also include inflammatory bowel disease or kidney disease. The severity of these problems varies much from person to person. Since health professionals cannot easily diagnose HPS with usual types of blood tests, it is important to promote awareness about the syndrome.

Although HPS has been studied most frequently among Puerto Ricans, the syndrome occurs all over the world. It was first recognized in two Czechoslovakian individuals in 1959. It should be suspected in any child with albinism who shows easy bruising or unusual bleeding, such as repeated nosebleeds.

The color of skin and hair varies in HPS and may resemble any other type of albinism. Some with HPS may have white or creamy skin as in type 1 albinism. Others may have sun freckling and yellow or light brown hair. A few with HPS may have dark brown hair and lightly pigmented skin, so that they appear to have ocular albinism.

The eye color varies from blue to brown. The eye problems are similar to those in other types of albinism:

- nystagmus— involuntary (side-to-side) movement of the eyes,
- strabismus— (“crossed” or “wall” eyes),
- sensitivity to glare,
- decreased acuity— 20/60 to 20/400.

Bleeding Problems

The bleeding problems of HPS result from a malfunction of platelets, the tiny blood cells that

clump together to plug up damaged blood vessels in cuts, scrapes, and bruises. The platelets lack dense bodies, which are tiny storehouses of the chemicals which platelets need to stay clumped together.

Often the bleeding tendency in HPS is mild, and easy bruising or nosebleeds may not lead to the diagnosis. Sometimes the bleeding can be severe, such as with surgery or childbirth. The use of aspirin and a wide variety of aspirin-related drugs can make the bleeding problem worse.

The usual tests which physicians use to diagnose coagulation (clotting) problems usually will show normal results in HPS. These tests which may be normal include the prothrombin time (PT), partial thromboplastin time (PTT), and platelet count. A bleeding time test is usually prolonged but may be normal.

Some individuals with HPS who have more severe bleeding problems have a second problem; they also have less of a blood clotting substance called von Willebrand factor. If von Willebrand factor is less than 70%, the risk of bleeding is high. Von Willebrand factor can be measured “with a blood test done in some hospital labs.

The diagnosis of HPS is made by a test not done in usual clinic, hospital, or reference labs. In this test, platelets from a blood sample are examined with an electron microscope. In HPS the platelets contain few or no dense bodies. To find out how to get the test done, contact the HPS Network (address below).

Lung Disease

HPS also includes lung disease. Many with HPS have pulmonary fibrosis, which means that the tissue of the lung scars, and restricts the inflation of the lungs. The lung problems tend to

worsen with time. Spirometry, which involves measuring air flow as a person blows into a machine, may show a problem before any chest x-ray changes appear.

Inflammatory Bowel Disease

Another problem in Hermansky-Pudlak Syndrome is inflammation of the intestines, which may cause bloody diarrhea or abdominal pain. The bowel problems in HPS are similar to those seen in a common chronic disease, Crohn's disease. In some cases, blood loss from the diarrhea can become severe enough to require blood transfusions or surgery.

The cause of the lung and intestinal problems may have something to do with a "ceroid" or yellowish material which is found in many different organs of people with HPS. Research has not yet found the exact nature of this material. Newer research has identified several genes for HPS, but scientists believe that more genes are yet to be found. The defect in HPS seems to involve Intracellular vesicular trafficking.

Severity Varies

One of the most troubling aspects of HPS is that its course is unpredictable. The severity of bleeding problems varies much in HPS, from minor bruising to life-threatening hemorrhage. Some children have had bleeding problems from minor surgery such as placement of ear tubes. The bowel disease usually does not develop until adolescence, although in several cases it has appeared in infants. The lung disease may not develop until adulthood.

Since its severity and onset vary so much, and since it is uncommon enough that most physicians have not heard of it, people with albinism need to know about HPS. Children with albinism should not take aspirin and products that effect platelet function unless it is certain that they do not have HPS. If a child with albinism shows any easy

bruising or colitis, or is to undergo surgery with risk of bleeding, it is important to inform health professionals about the possibility of the syndrome and direct them to sources of information about it.

To contact other families who have dealt with Hermansky-Pudlak Syndrome, contact:

Donna Appell
The HPS Network
1 South Road Oyster Bay, NY 11771-1905
Phone: 1800-789-9477 Fax: 516-922-4022
Toll free U.S.: 800-789-9HPS (9477)
e-mail: donna_appell@albinism.org

Articles for professionals:

Schinella RA, Greco MA, Colbert BL, Denmark WL, Cox RP: Hermansky-Pudlak syndrome with granulomatous colitis. *Ann Int Med* 1980, 92:20-3.

Mahadeo R, Markowitz J, Fisher S, Daum F: Hermansky-Pudlak syndrome with granulomatous colitis in children. *J. Peds* 1991, 904-906.

King RA, Hearing VJ, Creel DJ, Oetting WS: Albinism, 4353-4393, in Scriver, Charles R. et al, *The Metabolic and Molecular Bases of Inherited Disease*, 7th ed., McGraw Hill Inc. Health Professions Division, New York 1995. This textbook is available in most medical libraries.

Technical updates are accessible via internet from *Online Mendelian Inheritance in Man*, address: <http://www3.ncbi.nlm.nih.gov/Omim/searchomim.html>

Shotelersuk V, Gahl W: Minireview Hermansky-Pudlak Syndrome: Models for Intracellular Vesicle Formation. *Molecular Genetics and Metabolism* 1998, 65:85-96.

-Jim Haefemeyer MD, MS, Chair, NOAH Board of Scientific Advisors

--Reviewed by Richard King, MD, PhD, Director, International Albinism Center, Director of the Division of Genetics, Department of Medicine, University of Minnesota, and the NOAH Albinism Awareness Committee.

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PO Box 959, East Hampstead, NH 03826-0959, USA

Telephone: 603 887-2310 / 800 473-2310

Internet: <http://www.albinism.org> E-mail: info@albinism.org