



Hermansky-Pudlak Syndrome

What is Hermansky-Pudlak Syndrome?

Hermansky-Pudlak Syndrome

is characterized by:

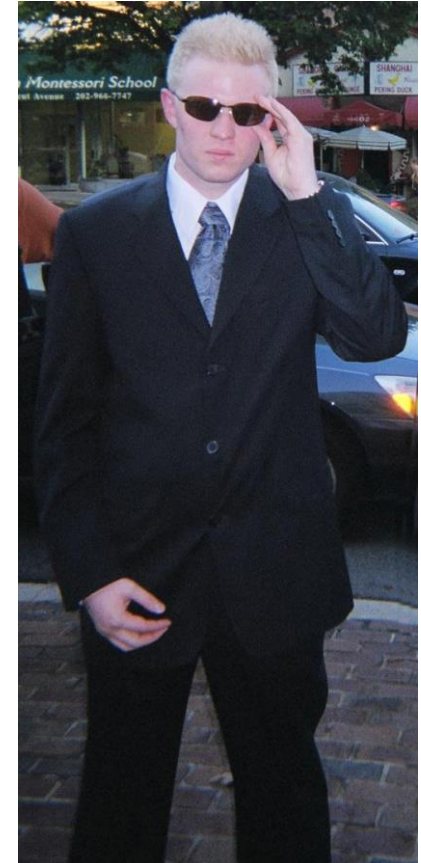
- Albinism
- Vision impairment
- A bleeding disorder
- Inflammatory Bowel Disease
- Pulmonary Fibrosis or lung scarring
- Heart and kidney problems in some advanced cases

Albinism

Albinism is a decrease in pigmentation:

- Some individuals lack pigment in their hair, skin and eyes
- Others may have brown hair and brown eyes but their eyes still lack pigment causing transillumination

Who has HPS?



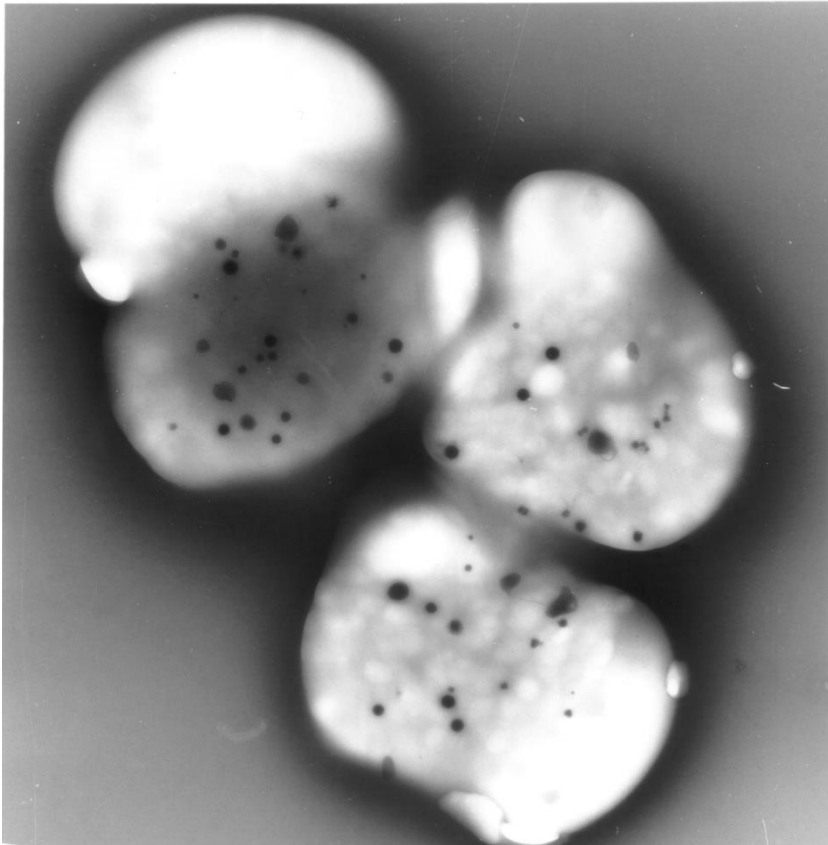
Answer: **All of them!**

Bleeding disorder

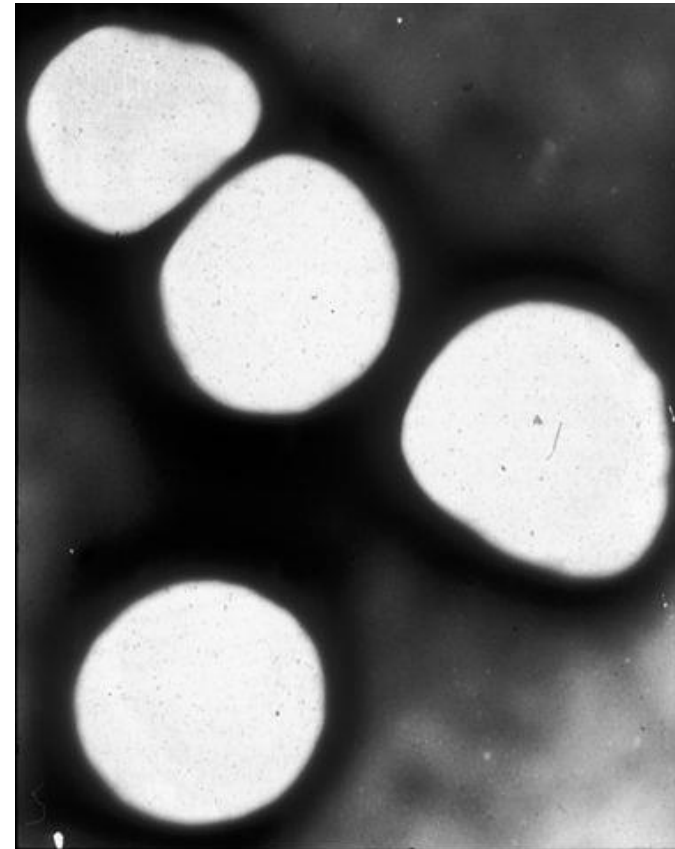
A platelet defect caused by abnormal formation and structure that can result in:

- Easy bruising
- Nosebleeds
- Prolonged menstruation
- Difficulty clotting during surgical and dental procedures

Bleeding disorder: platelets lack dense bodies



Chocolate Chip Cookies
Normal platelets



Butter Cookies
HPS platelets

Bleeding disorder: warnings

- It's important for those with HPS to get diagnosed so in the event of injury or surgery they can receive proper treatment.
- It's important for those with HPS to avoid aspirin, products containing aspirin, ibuprofen (Motrin), naproxen (Aleve) and any other products, which may compromise platelet function.

Inflammatory bowel disease

- Incidence: 15 to 20% of people with HPS develop digestive problems
- Can occur anywhere in the digestive track
- Is very much like Crohn's disease
- Symptoms include:
 - diarrhea
 - blood in stool
 - cramps
 - weight loss

Inflammatory bowel disease

- Often strong and expensive medications are required to control colitis.
- In severe cases, it may be necessary to remove part of the large intestines.
- It's important to distinguish between the severity of the bleeding and the severity of the bowel disease.

Pulmonary Fibrosis

- Is a hardening or scarring of the lungs
- HPS types 1,2 and 4 develop pulmonary fibrosis
- Some of the symptoms are:
 - shortness of breath
 - abnormal fatigue with exercise and exertion
- Frequently misdiagnosed for asthma
- Usually fatal in the fourth or fifth decade

Pulmonary Fibrosis: Treatment

- Presently there is no cure for Pulmonary Fibrosis
- There are antifibrotic medications that can be used to slow down the lung scarring
- HPS Network has participated in 2 drug trials with the antifibrotic drug pirfenidone (Esbriet)
- HPS members with severe lung disease have also been able to obtain a lung transplant

Over 30 of our members has successful Lung Transplants as the
only treatment for the lung diseases
Here are some of them!!



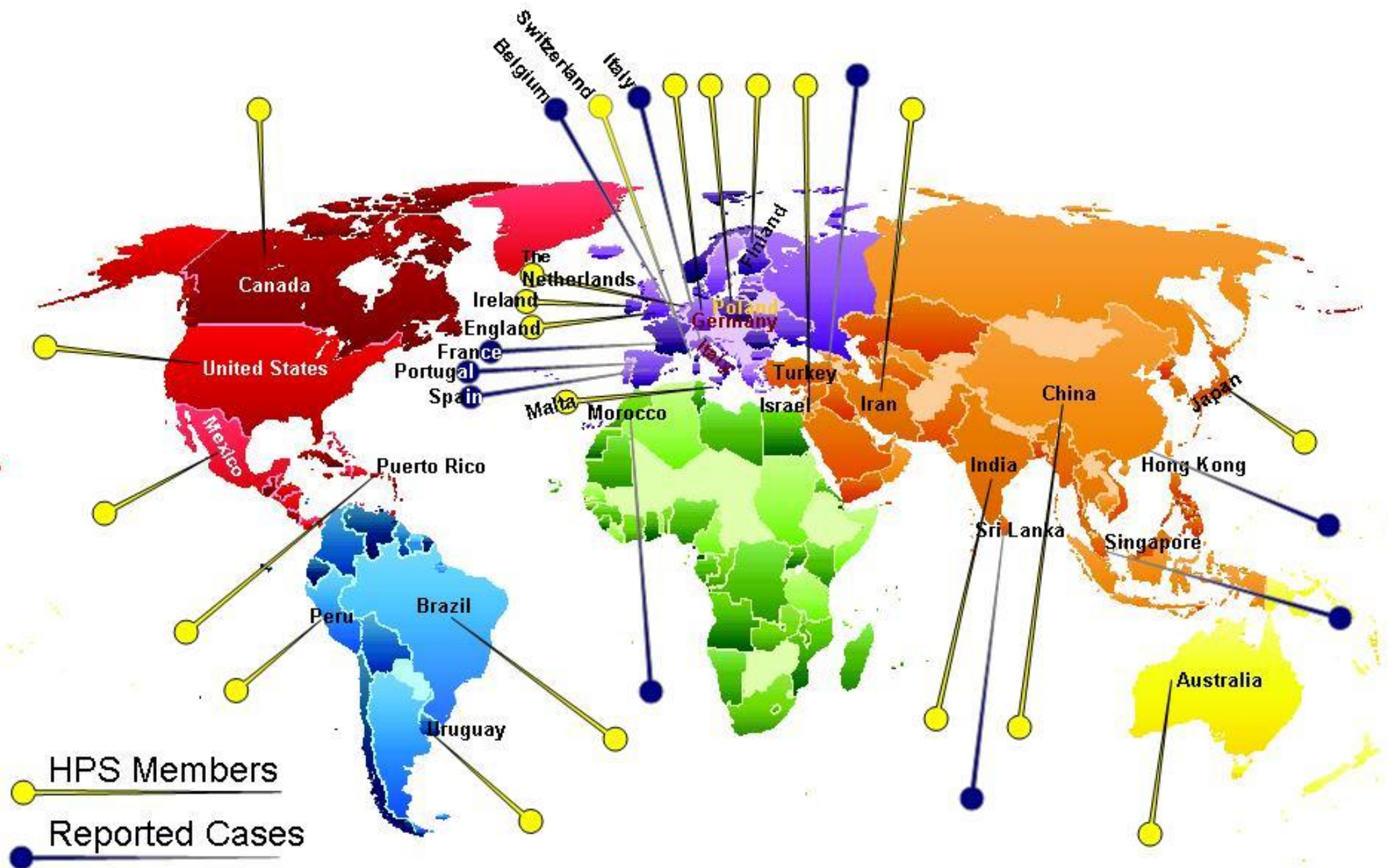
Genetics of HPS

- Usually a recessive gene that is carried by both parents
- Currently there are ten different gene types of HPS
- HPS types 1, 2 and 4 develop pulmonary fibrosis
- HPS 2 is known to be prone to infections

Statistics: General population

- Carrier Rate: 1:350 – 1:500
- The chance that two carriers meet and have children: 1:125,000
- Rate of babies born with HPS in the general population is 1:500,000 – 1:1,000,000
- The incidence of HPS₁: 1:1600 due to founders effect
- We really do not have accurate statistics of the rate of HPS as a diagnosis protocol is not available around the world

Hermansky-Pudlak Syndrome – Around the World



These countries make up those registered on the HPS database

■	Algeria	1	■	Ireland	1
■	Australia	6	■	Israel	1
■	Belgium	1	■	Italy	1
■	Bolivia	1	■	Japan	1
■	Brazil	1	■	Malta	1
■	Canada	10	■	Mexico	2
■	China	1	■	Netherlands	4
■	Ecuador	1	■	Pakistan	1
■	England	10	■	Peru	1
■	Finland	2	■	Poland	2
■	France	2	■	Puerto Rico	
■	Georgia	1	■	Portugal	1
■	Germany	6	■	Qatar	1
■	Holland	2	■	South Arabia	1
■	Hungary	2	■	Switzerland	1
■	India	3	■	Turkey	1
■	Iran	1	■	United States	
			■	Uruguay	1

Cases reported in the medical literature

- Argentina
- Cuba
- El Salvador
- Hong Kong
- Morocco
- Norway
- Argentina
- Bolivia
- Singapore
- Spain
- Singapore
- Spain
- Sri Lanka
- Ukraine

We are aware that there are individuals with HPS in this countries and more individuals in the countries where we have members registered with us, but we are not able to count them in our registry unless they are registered with the HPS Network.

What is the HPS Network?



The HPS Network

- Is a 501(c)(3) non-profit founded in 1992 and incorporated in 1995
- Maintains a patient registry
- Provides support and education to families
- Helps families obtain a diagnosis
- Educates medical professionals
- Promotes and funds HPS research
- Facilitates an online community
- Hosts annual conferences



Global Albinism Alliance
Paris , France
January 24-27 2020

Activities of our region

- We offer two conferences every year, One in NY and one in PR. We also participate in National and International medical conferences, like the American Thoracic Society as well as National Organizations of Rare Diseases
- We host the meeting of the minds, where we gather over 40 doctors, scientist and researchers interested in studying HPS and hopefully find a cure for the conditions associated with this type of albinism. This meeting is the only of its kind for HPS and the requirement is that speakers must agree to discuss cutting edge research and unpublished data.
- We keep an international CONTACT REGISTRY with people diagnosed with HPS as well as reported cases of HPS around the world.
- We spend a lot of time responding to emails and calls regarding diagnosis and is the nurse's office of the albinism community. Two nurses are on the other end of the phone. Often, we answer a lot of questions about the inheritance of albinism. We do not ask for dues.
- We promote collaboration between researchers and our HPS community to develop research, studies that will bring us closer to a cure, We support our membership and educate them in the process of scientific trials, medication and self-care so they can fully participate in research if they choose to. We helped them develop an Individualized Research Plan to help them sort out what their comfort of involvement with research is as well as creates a data base of our members availability for new research trials.
- We do outreach in our communities as well as keep a presence internationally through our social media and, Website and Facebook page.
- We develop the TIPS program (Transplant Information and Patient Support) to help individuals and their family navigate the transplant process and provide them with tools to develop their support system and understand the process.
- We sponsor in conjunction with the Lighthouse Guild the parent support call where I facilitate a call in English the second Tuesday of each month and a call in Spanish, every Monday. We offer parent information about HPS, standard of care, bring expert to answer parents' questions as well as provide the parent s a forum to express concerns or share their experiences of rising a child with HPS.

Challenges of the region

- Our biggest challenge is appropriate diagnosis and fighting the fallacy that HPS is only a problem in the Puerto Rican Community. HPS is present globally and should be made aware to all the albinism community as ignoring the facts can end up in dier consequences.
- Another challenge is to provide our families genetically identified with HPS type 1, 2 and 4 constant hope and emotional support to combat the stress of knowing that without medical intervention they may die. Two years agho we lost 14 of our members, we are loosing a generation.

Region's Opportunities

- We are looking forward to creating a network of international researchers as we continue to gather our global community

Best Practices

- We support Albinism awareness day as well as Rare Disease Day
- We launched last year HPS AWARENESS DAY on April 6Th (the day our organization was registered as a non for profit organization.)
- We work on continuous collaboration with doctors, researchers and scholars in the US and around the world to develop treatment and ultimately a cure for the medical issues associated to HPS and other syndromes of albinism.
- We work in close collaboration with NOAH as they address all the psychosocial and labor issues relevant to all the PWAs. The HPS Network reliably provides the medical and safety information to support comprehensive educational and healthcare tools for families to understand the implications of HPS in our albinism community.
- We encourage self-advocacy and are a credible source for resources for medical professionals and the community at large.
- We are the leading organization that has a contact registry of individuals with HPS which we hope to expand through a global collaboration.

Conferences and Outreach

- Annual Conferences:
 - New York Conference
 - Puerto Rico Conference
- Contributions to Sponsorships
 - Gordon Research Seminars
 - Federation of American Societies for Experimental Biology
- Outreach and Professional Collaborations:
 - American Society of Human Genetics
 - American Thoracic Association
 - CHEST
 - American Society of Genetic Counselors
 - Digestive Disease Week
 - International Society on Thrombosis and Haemostasis Congress

Annual Conferences



New York

Puerto Rico







Funds research

Our most recent grant was in partnership with the American Thoracic Society



Promotes awareness



Medical conferences



Advocates on Capitol Hill

The Network has advocated for research funding, the Genetic Information Non-Discrimination Act, medical coverage for pulmonary rehabilitation and the freedom for those with oxygen to travel, uninhibited, on airplanes.



The NIH Clinical Center

The HPS Network has participated in:

- Research
- Natural history study
- Multi-drug trial for severe PF
- Two drug trials for Pirfenidone



HPS Centers of Excellence

- Vanderbilt University Medical Center-
Dr. Lisa Young
- Brigham and Women's Hospital- Dr. El-
Chemaly
- Loyola University Medical Center- Dr. Daniel
Dilling

Lung Disease Clinic

- Mayaguez Medical Center, PR- Dr. Jesse
Roman and Dr. Rosa Roman -Medical Director

Why spend a lot of money researching something so rare?

- HPS is teaching science about:
 - skin conditions
 - visual impairments
 - platelet disorders
 - inflammatory bowel diseases
 - pulmonary fibrosis
- Researching the rare will help cure the common
- Millions may one day benefit from our research!

India



India



Puerto Rico



Japan



United
Kingdom



United States



Puerto Rico



Contact Us

HPS Network
One South Road
Oyster Bay, New York 11771-1905

Telephone: 1 (800) 789-9HPS
Fax: 1 (516) 624-0640

e-mail address: info@hpsnetwork.org

Web site: www.hpsnetwork.org