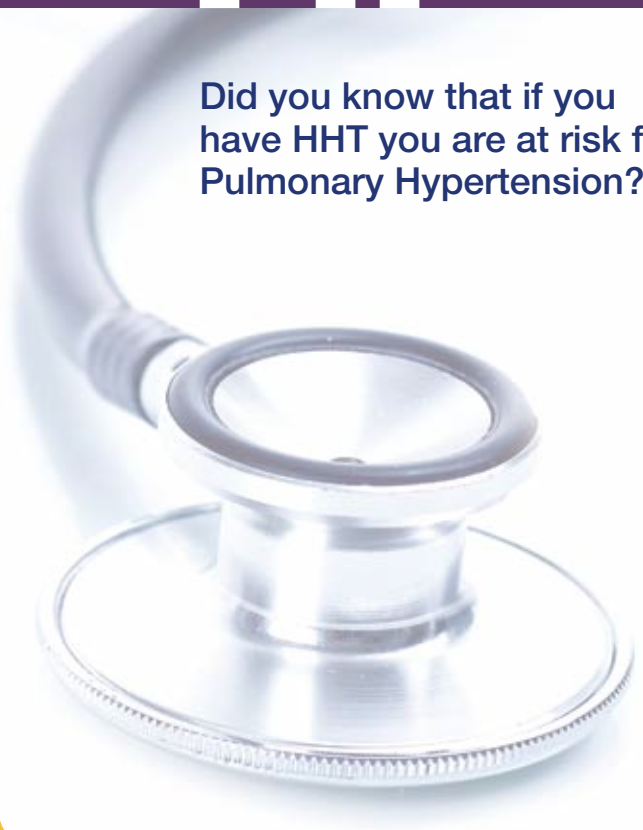
Hereditary Hemorrhagic Telangectasia (HHT)

& PH Pulmonary Hypertension



Did you know that if you have HHT you are at risk for Pulmonary Hypertension?

Pulmonary Hypertension Association
Empowered by hope

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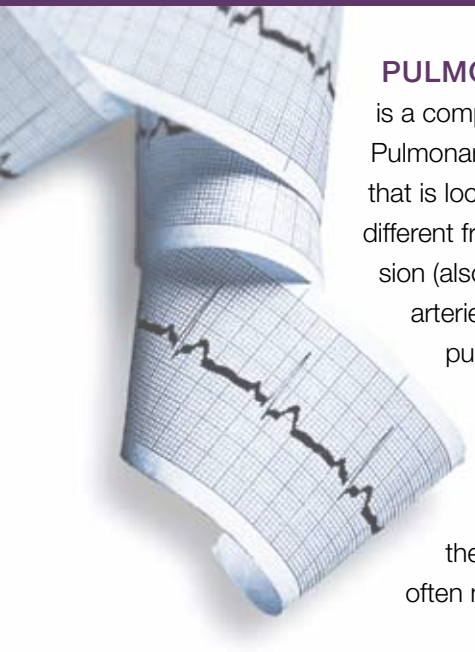
About Pulmonary Hypertension



PULMONARY HYPERTENSION, OR PH, is a complex and often misunderstood disease. Pulmonary hypertension means high blood pressure that is located in the lungs. Pulmonary hypertension is different from regular hypertension. In regular hypertension (also known as high blood pressure), a person's arteries throughout the body are constricted. In pulmonary hypertension, the blood vessels in the lungs become narrowed and the heart has to work harder to pump blood through them. PH can also be caused by multiple blood clots, diseases that damage the lung, or heart failure. Early symptoms are often non-specific and diagnosis is often delayed.

Pulmonary hypertension affects people of all ages and ethnic backgrounds. The most common symptoms are shortness of breath with physical activity, fatigue, lightheadedness and sometimes fainting. Because these symptoms can be caused by any number of other problems, diagnosing a case of pulmonary hypertension can be difficult and may require a specialist. Once pulmonary hypertension is diagnosed, however, treatment can begin immediately.

One form of PH is called pulmonary *arterial* hypertension (PAH). In PAH, the blood vessels that carry blood to the lungs (known as the pulmonary arteries) are narrowed, thickened and stiff. Because of this, the pressure in those arteries is abnormally high. This means that the right side of the heart has to pump much harder to move blood. There are several types of PAH. In what is known as idiopathic PAH (IPAH), the cause of the PAH is never known. In what is known as familial PAH (FPAH), the disease is inherited, meaning another member or members of the patient's family was diagnosed with pulmonary hypertension. PAH can also be associated with other medical conditions such as connective tissue diseases (scleroderma and lupus, for example), chronic liver disease, congenital heart disease, sickle cell disease and HIV infection. Finally, PAH can be associated with past or present drug use, such as methamphetamines or certain diet pills.

Some patients who are diagnosed with PH also suffer from other specific problems as well. These other problems might include left-sided heart disease (for example, people who have had heart attacks, have hypertension or heart valve disease), chronic obstructive pulmonary disease (COPD) and emphysema, and chronic thromboembolic disease (chronic blood clots in the lungs).

PAH is a serious condition, and without treatment, symptoms can only become worse, leading to heart failure and even death. Therefore, it is essential that patients follow the advice of their doctor. PAH, like PH itself, is rare and its signs and symptoms are vague and often confused with other disorders. Proper diagnosis and therapy are essential. Increasing public awareness of PAH (and PH) will help patients seek treatment earlier, which can lead to better long-term health. While no cure has yet been found for PH, increased research has resulted in new and better medications and therapies that allow PH patients to live longer, fuller lives with far less interference from the disease. Even more promising research is being conducted every day, and advances are made every year.

PH in Association with Hereditary Hemorrhagic Telangectasia, or HHT

My doctor says I have Hereditary Hemorrhagic Telangectasia. What exactly does that mean?

Hereditary Hemorrhagic Telangectasia (HHT) is a genetic disorder that causes abnormalities of blood vessels, usually resulting in excessive bleeding (known as hemorrhaging). In order to understand HHT, you must first understand the way blood is pumped to and from the heart. Blood vessels are hollow structures that transport blood throughout the body. There are two types of blood vessels: arteries and veins. Arteries carry blood under high pressure *away from the heart* to all other areas of the body. Blood then passes through capillaries, where oxygen, nutrients and waste products can be exchanged. Veins carry blood that should be under low pressure *back to the heart*. An artery does not usually connect directly to a vein.

A person with HHT has a tendency to form blood vessels that lack capillaries between the arteries and veins, meaning that arterial blood under high pressure flows directly into a vein without first going through the very small capillaries. The site of this abnormal connection tends to be fragile and can rupture and result in bleeding. This type of abnormal blood vessel is called a telangiectasia if it involves small blood vessels. It is called an arteriovenous malformation (AVM) if it involves large blood vessels. Telangiectases usually occur on the surface of the body, such as the skin and the mucous membranes that line the nose and the gastrointestinal tract. AVMs are found in the lungs, liver and central nervous system.

My doctor has asked me whether I feel lightheaded, out of breath or faint, and has said these might be symptoms of pulmonary hypertension. How does this relate to my HHT?

About 15 to 20 percent of people with HHT have at least mildly elevated pulmonary artery pressures, which means they either have or are developing PH. HHT patients can develop PH in two ways and the differences are significant.

In HHT-associated pulmonary arterial hypertension, abnormal blood flow through the blood vessels in the lungs causes elevated blood pressure. While it is not known exactly how many people have HHT-associated PAH, research has identified one affected member in 15 percent of families with a form of HHT known as HHT2. In another type of HHT, known as HHT1, HHT-associated PAH appears to be much less common.

More commonly, PH can develop in HHT patients as a result of increased blood flow from the heart, a condition called high cardiac output state. A high cardiac output state may be the result of several things, and a doctor may be able to determine the cause. Treatment will vary depending on what type of PH you have.

How do doctors test for this disease?

It is often difficult to detect PAH in HHT patients. In fact, some of the symptoms of PAH (fatigue, difficulty breathing and difficulty during physical exertion) are already found in people with HHT. To make things even more confusing, these symptoms are often seen in HHT patients as side effects of other health problems including heart failure, anemia and liver problems. Therefore, in many cases, a doctor must strongly suspect PAH, or it may go overlooked.

An echocardiogram can test for abnormally high blood flow in the lungs of HHT patients, and also can screen for PH. In addition, the echocardiogram provides important information about the heart, including its size and function. This test can also help uncover underlying problems the patient may be facing.

If PH is suspected in an HHT patient or diagnosed through an echocardiogram, another test, a right heart catheterization, can help doctors confirm it, and can also help to diagnose the exact type of PH the patient is suffering from.

What can be done for me?

Treatment is available; however, in order for a doctor to accurately treat HHT-associated PH, he or she must first diagnose the specific type of PH the patient has. If HHT-associated PAH is diagnosed, PAH specific therapies can be carefully prescribed, but these medications can also aggravate bleeding tendencies that are already present in HHT patients. As a result, HHT patients with PAH should seek out a specialist at a PH treatment center.

PHA Resources

The Pulmonary Hypertension Association (PHA) was founded by and for PH patients. The organization has led the way in bringing pulmonary hypertension into the national and international consciousness. PHA is constantly increasing its services to the medical community through educational programming, membership sections for medical professionals, and much more:

Website:

PHA's website is a comprehensive source of information for patients, caregivers and medical professionals. Please visit us at **www.PHAssociation.org**.

Find a Doctor:

The "Find a Doctor" section of PHA's website allows patients and referring physicians to search for PH-treating physicians by state at: **www.PHAssociation.org/Patients/FindADoctor**. While PHA does not endorse any of these physicians, PHA strongly recommends that all PH patients see a PH specialist who will be able to provide them with the best PH care.

Pulmonary Hypertension: A Patient's Survival Guide:

This extraordinary 300+ page book was written by a patient and medically reviewed. It presents the illness in a very human and readable way. It is available from PHA at minimal cost, at a discount for PHA members, and one complimentary copy is provided to members of PHA's professional membership bodies. *The Survival Guide* is available for purchase online at **www.PHAssociation.org/SurvivalGuide**.

Online information about PH:

For information on PH diagnosis, symptoms, treatments and more, visit **www.PHAssociation.org/Patients/AboutPH**.

Support Groups:

From the first support group started in 1990 around a kitchen table in Florida, PHA grew to 45 in 2001, and to over 215 in 2010. In many places, patients have the opportunity to meet, learn from, and find common understanding with others in similar circumstances. Find a support group in your area at **www.PHAssociation.org/LocalSupportGroups**.

About the Pulmonary Hypertension Association

The mission of the Pulmonary Hypertension Association is to find ways to prevent and cure pulmonary hypertension, and to provide hope for the pulmonary hypertension community through support, education, advocacy and awareness. PHA's members stand as part of a community that is fighting back against this terrible illness.

PHA fulfills its mission through:

- Funding for research
- Quarterly medical journal *Advances in Pulmonary Hypertension*
- PHA Online University offering free CME credits and the latest information on pulmonary hypertension **(www.PHAOnlineUniv.org)**
- Professional membership sections:
  - PH Clinicians and Researchers (PHCR)—for physicians and doctorate-level researchers
  - PH Resource Network—for nurses and allied health professionals
- Educational conferences and materials for medical professionals and patients
- 300+ page patient's survival guide
- PH patient support groups
- Quarterly newsletter *Pathlight*
- Advocacy and awareness campaigns
- Toll-free Patient-to-Patient Helpline (1-800-748-7274)
- PHA website with PH discussion boards, email groups and online support chats **(www.PHAssociation.org/ConnectOnline)**

More Information on Hereditary Hemorrhagic Telangectasia (HHT)

HHT Foundation International
www.hht.org

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