Did you know that if you have HHT, you are at risk for pulmonary hypertension?

Hereditary Hemorrhagic Telangiectasia (HHT) & Pulmonary Hypertension

www.PHAssociation.org
PH 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 medical problems, diagnosis is often delayed. Identifying a case of PH can be difficult and may require a specialist. Once the type of PH is diagnosed, however, treatment can begin immediately.

One form of PH is called pulmonary arterial hypertension (PAH). PAH is a complex, progressive type of PH where the high blood pressure in the lungs occurs because tiny blood vessels that carry blood through the lungs (pulmonary arteries) are narrowed, thickened and stiff. As PAH advances, the heart may lose its ability to pump enough blood through the lungs to meet the needs of the body.

There are several types of PAH. Idiopathic PAH (IPAH) means that the patient develops PAH without any other obvious medical problem leading to high blood pressures in the lung. Heritable PAH (HPAH) comes from abnormal genes that cause PAH. Heritable PAH may be passed on to some members of your family. PAH can be associated with other medical conditions such as connective tissue diseases (scleroderma and
lupus for example), chronic liver disease, congenital heart disease, or HIV infection. Finally PAH can be associated with past or present drug use, such as methamphetamines or certain diet pills. It is not known exactly how these medical problems or drugs cause PAH.

PAH is a serious condition, and without treatment, symptoms can only become worse, leading to heart failure and even death. Proper diagnosis and therapy from a doctor who understands PAH is essential. Every patient is different. The choice of treatment is based on how sick a patient is and the risks and benefits of any particular therapy. Regardless of risk, is it important that patients and their healthcare providers engage in frequent follow-ups with ongoing discussions about the management of their condition. Current guidelines suggest that changes in therapy should be considered for patients not reaching their treatment goals.

While no cure has yet been found for PAH, increased research has resulted in treatments that allow patients to live longer, fuller lives with far less interference from the disease. Even more promising research is being conducted every day that is not only advancing our understanding of the PAH disease but also potentially identifying new treatment options for patients in the future.
My doctor says I have hereditary hemorrhagic telangiectasia-associated PH. What exactly does that mean?

Hereditary hemorrhagic telangiectasia-associated PH (HHT-associated PH) is a genetic disorder that causes abnormalities of blood vessels, usually resulting in excessive bleeding (known as hemorrhaging).

In order to understand HHT-associated PH, 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-associated PH 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-associated PH?

About 15 to 20 percent of people with HHT-associated PH have at least mildly elevated pulmonary artery pressures, which means they either have or are developing PH. HHT-associated PH patients can develop PH in two ways, and the differences are significant. In HHT-associated pulmonary arterial hypertension (PAH), 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 often, 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 factors, 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 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. Catherization can also help to diagnose the exact type of PH the patient is experiencing.

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.
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](http://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](http://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 is medically reviewed and updated annually. It presents the illness in a very human and readable way, covering a wealth of topics like the mechanics of PH, the latest treatments, patient care and lifestyle issues. PHA members receive a discount on this resource. The *Survival Guide* is available for purchase as a paperback and an e-book at: [www.PHAssociation.org/SurvivalGuide](http://www.PHAssociation.org/SurvivalGuide).

**Online information about PH:**
For information on PH diagnosis, symptoms, treatments and more, visit [www.PHAssociation.org/Patients/AboutPH](http://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 groups in 2001 and to more than 245 in 2013. 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](http://www.PHAssociation.org/LocalSupportGroups).
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, research, advocacy and awareness. PHA’s members form 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](http://www.PHAOnlineUniv.org))
- Professional membership sections:  
  - PH Clinicians and Researchers (PHCR)—for physicians and doctorate-level researchers  
  - PH Professional Network—for nurses and allied health professionals
- Educational conferences and materials for medical professionals and patients
- A wealth of information in the *Survival Guide*
- PH patient support groups
- Quarterly newsletter *Pathlight*
- Advocacy and awareness campaigns
- Toll-free Patient-to-Patient Support Line (1-800-748-7274)
- PHA website with PH discussion boards, email groups and online support chats ([www.PHAssociation.org/ConnectOnline](http://www.PHAssociation.org/ConnectOnline))

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**More Information on Hereditary Hemorrhagic Telangiectasia (HHT)**

HHT Foundation International  
[www.hht.org](http://www.hht.org)