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# Pulmonary Hypertension & HEREDITARY HEMORRHAGIC TELANGIECTASIA

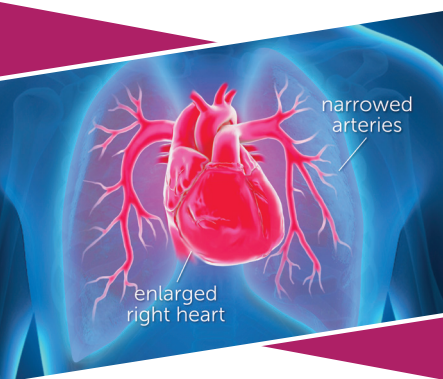


Pulmonary Hypertension Association  
Empowered by hope

# ABOUT PULMONARY HYPERTENSION

Pulmonary hypertension (PH) is a complex and often misunderstood disease. The term PH simply means high blood pressure in the arteries of the lungs. In regular hypertension — also known as high blood pressure or systemic hypertension, which you can check with a blood pressure cuff — the pressure in the arteries of the entire body is higher than it should be. In PH, the blood vessels specifically in the lungs are affected. They can become stiff and narrow, and the right side of the heart must work harder to pump blood through them. There are five groups of PH based on different causes.

Each form of PH is different, so it is important for newly diagnosed patients to find a PH specialist who can accurately pinpoint what is causing their PH. They then can develop a treatment plan for that specific type as soon as possible after a confirmed diagnosis. Every individual with PH is different, and new research with the potential to improve the outlook for people living with this disease is conducted on a continual basis. Once in the care of a PH-treating health care team and on appropriate therapy, individuals with PH can live for many years.



*PH, or high blood pressure in the lungs, frequently results from a narrowing of the small blood vessels in the lungs, which leads to a larger right side of the heart.*

*Normal mean pulmonary artery pressure is between **8-20 mmHg** at rest. Pulmonary hypertension is defined as a resting mean pulmonary artery pressure at or above **25 mmHg**.*





## HEREDITARY HEMORRHAGIC TELANGIECTASIA-ASSOCIATED PH

Hereditary hemorrhagic telangiectasia (HHT) is a genetic disorder that causes abnormalities of blood vessels, usually resulting in excessive bleeding (known as hemorrhaging). In order to understand how PH can develop, it is important to understand the way blood is pumped to and from the heart. Blood vessels transport blood throughout the body. There are three types of blood vessels: arteries, veins and capillaries. Arteries carry blood under high pressure away from the heart to other areas of the body. Blood then passes through capillaries — small blood vessels where oxygen, carbon dioxide, nutrients and waste products can be exchanged. Veins carry blood under low pressure back to the heart. An artery does not usually connect directly to a vein without first passing through capillaries.

In HHT, the capillaries can be lacking in between the arteries and veins, so that arterial blood under high pressure flows directly into a vein. 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. Telangiectasias 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.

About 13 percent of people with HHT have at least mildly elevated pulmonary artery pressures.<sup>2</sup> HHT-associated PH patients can develop PH in two ways and the differences are significant. These two forms of HHT-associated PH are managed very differently.

Most commonly, PH develops in HHT patients as a result of increased blood return to the heart due to large liver AVMs, which leads to a condition known as high-output heart failure (HOHF). In this situation, the arteries of the lungs are dilated (widened) and resistance to blood flow is low, but pressures still are mildly elevated due to increased blood flow through the lungs and high pressures downstream from the lungs in the left side of the heart. Anemia, which is common in HHT, will further worsen this type of heart failure and PH.

In HHT-associated World Health Organization Group 1 PH (pulmonary arterial hypertension, or PAH), a heritable form of PAH, pressures in the arteries rise significantly due to increased resistance to blood flow through the lungs because of abnormally narrowed blood vessels. HHT-associated PAH is quite rare and estimated to occur in less than 1 percent of HHT patients, primarily in individuals with mutations in the ACVRL1 gene (formerly known as ALK1).<sup>3,4</sup>

<sup>2</sup> Olivieri C, et al. Genet Med. 2006;8(3):183-90.

<sup>3</sup> Vorselaars V, et al. Respiration. 2017;94(3):242-250.

<sup>4</sup> Berg JN, et al. Am J Hum Genet. 1997;61(1):60-7.

# DIAGNOSING HHT-ASSOCIATED PH

It often is difficult to detect PH in HHT patients. In fact, some of the symptoms of PH (fatigue and difficulty breathing, and especially with physical exertion) often are found in people with HHT due to other health conditions, including heart failure, anemia and liver problems. Therefore, a doctor must strongly suspect PH in many cases or it may go overlooked. An echocardiogram (lung ultrasound) can test for abnormally high blood flow in the lungs and also can be used to screen for PH. In addition, the echocardiogram provides important information about the heart, including its size and function. If PH is suspected in an HHT patient or evidenced after an echocardiogram, a right heart catheterization will help doctors confirm the diagnosis and determine which type of PH someone has and how severe it is.



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# TREATMENT OPTIONS FOR HHT-ASSOCIATED PH

Treatment is available for HHT-associated PH. However, in order for doctors to accurately treat HHT-associated PH, they first must diagnose the specific type of PH. When PH is due to HOHF, management should focus on treating heart failure with diuretics (water pills), correcting anemia, and possibly using anti-angiogenic therapy (to prevent the formation of new blood vessels) administered by an HHT expert. PH-specific therapies can be used carefully, but these medications can aggravate bleeding, which already is a problem for many HHT patients. As a result, individuals with HHT-associated PH should seek out specialist care from PH centers that will collaborate with HHT experts.



# ABOUT THE PULMONARY HYPERTENSION ASSOCIATION

The Pulmonary Hypertension Association (PHA) is a community of individuals with PH, caregivers, families, medical professionals and researchers who work together to empower people battling the disease, while supporting research toward improved treatments. PHA was the first organization in the world dedicated to providing comprehensive PH patient and caregiver support, medical education, research and services that improve patients' quality of life.

*PHA's mission is to extend  
and improve the lives of  
those affected by PH.*

*PHA's vision is a  
world without PH,  
empowered by hope.*

PHA facilitates more than 200 active support groups around the country and serves as a model for more than 85 international PH organizations. In addition to in-person support, PHA offers a telephone support line, an email mentorship program, educational patient programs and a variety of print and online publications. Every two years, PHA hosts its International PH Conference and Scientific Sessions, which brings together nearly 1,500 individuals with PH, caregivers and medical professionals to share information on the latest research, treatments and lifestyle coping tips.

PHA also has provided more than \$8 million for PH research and offers continuing education programs for medical professionals through the *PHA Online University* and an in-person CME program.

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