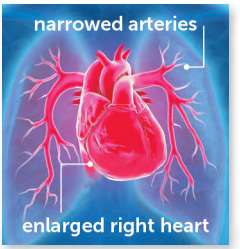
Pulmonary Hypertension and Hereditary Hemorrhagic Telangiectasia

PH&

PHA
Pulmonary Hypertension Association
Empowered by hope

ABOUT PULMONARY HYPERTENSION

Pulmonary hypertension is a complex and often misunderstood disease. PH sometimes is confused with systemic hypertension or "high blood pressure" that affects arteries throughout the body.



PH refers to pressure within the blood vessels of the lungs. The blood vessels can become stiff and narrow, which makes it more difficult for the right side of the heart to pump blood through them.

There are five types of PH based on different causes. Each form of PH is different, so it is important for newly diagnosed patients to find PH specialists who can pinpoint the cause of their PH. The specialist then develops a treatment plan specifically for the patient's type of PH.

Every person with PH is different, and scientists continually conduct new research to improve the outlook for people living with PH. With proper care and treatment, people with PH can live many years.

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 20 mmHg.

HHT-Associated PH

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

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 usually doesn't connect directly to a vein without first passing through capillaries.

With HHT, the capillaries can be missing between the arteries and veins, so arterial blood under high pressure flows directly into a vein. The site of the abnormal connection tends to be fragile and can rupture, causing bleeding.

The abnormal blood vessel is called a telangiectasia if it involves small blood vessels. It is called an arteriovenous malformation if it involves large blood vessels.

Telangiectasias usually occur on the body's surface, such as the skin and mucous membranes that line the nose and gastrointestinal tract. Arteriovenous malformations 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.

HHT-associated PH can develop 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 arteriovenous malformations. That leads to a condition known as high-output heart failure.

In this situation, the arteries of the lungs are dilated (widened), and resistance to blood flow is low. But pressures are mildly elevated because of 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.

HHT can sometimes cause a form of heritable pulmonary arterial hypertension. Abnormally narrowed blood vessels hinder blood flow through the lungs, causing pressure in the arteries to rise significantly.

HHT-associated PAH is quite rare and estimated to occur in less than 1 percent of HHT patients, primarily in people with mutations in the ACVRL1 gene (formerly known as ALK1).

Diagnosing PH

PH is often difficult to detect in people with HHT. In fact, some symptoms of PH (fatigue and difficulty breathing, especially with physical activity) often develop in people with HHT because of other health conditions. Those conditions include heart failure, anemia and liver problems. Unless a doctor strongly suspects PH, it might be overlooked.

Echocardiogram: An ultrasound of the heart that provides a rough estimate of the pressure in the lungs. It assesses the function of how blood flows through the heart by examining the right and left ventricles, valves, etc.

Pulmonary function tests: Non-invasive tests that measure how much air the lungs can hold, how much and how quickly someone can blow air, and how oxygen and carbon dioxide are exchanged between the air entering the lungs and the blood circulating through the lungs (diffusion capacity of carbon monoxide).

Ventilation-perfusion (V/Q) scan: Determines whether there are undissolved clots in the blood vessels of the lungs.

Natriuretic peptide tests: Measure specific proteins in a blood sample and help screen for heart failure. Tests of the proteins NT-proBNP (N-terminal pro b-type natriuretic peptide) and BNP (brain natriuretic peptide) are two examples. Elevated levels of these proteins can signify heart failure.

Right heart catheterization: The most accurate diagnostic test for PH. It measures the pressures and flow directly in the pulmonary artery. The invasive procedure usually takes place on an outpatient basis in the hospital. Doctors guide a small tube (catheter) through the right side of the heart and into the pulmonary artery to directly measure the pulmonary artery pressure.

In addition to directly measuring pressures in the heart and lungs, a right heart catheterization can determine whether patients have problems with a stiff left side of the heart. Left heart disease also can cause PH. In those cases, doctors refer patients to a cardiologist to treat the heart disease.

Treatment Options

Treatment is available for HHT-associated PH. However, to accurately treat HHT-associated PH, doctors first must determine the specific type of PH.

When PH is caused by high output heart failure, doctors should focus on:

- Treating heart failure with diuretics (water pills).
- Correcting anemia.

Possibly using anti-angiogenic therapy administered by an HHT expert to decrease the size of vascular liver malformations and slow the return of blood to the heart.

PAH-specific therapies can be used carefully for heritable PAH, but those medications can aggravate bleeding, a problem for many people with HHT. People with HHT-associated PH should seek specialist care from PH centers that will collaborate with HHT experts.

ADDITIONAL RESOURCES

Cure HTT | CureHHT.org

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ACKNOWLEDGMENTS

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About the Pulmonary Hypertension Association

Headquartered in Washington, D.C., the Pulmonary Hypertension Association is the oldest and largest nonprofit patient association dedicated to the pulmonary hypertension community. PHA's mission is to extend and improve the lives of those affected by PH.

PHA engages people with PH and their families, caregivers, health care providers and researchers, who work together to advocate for the PH community, provide support to patients, caregivers and families, offer up-to-date education and information on PH, improve quality patient care, and fund and promote research. For information, visit PHAssociation.org.

PHA's mission is to extend and improve the lives of those affected by pulmonary hypertension. PHA's vision is a world without PH, empowered by hope.

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