Did you know that if you have congenital heart disease, you are at risk for pulmonary hypertension?
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 meth-
amphetamines 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, it is 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.

**I have a congenital heart defect. How common is this?** Congenital heart defects affect slightly less than 1 percent of the population. Because of the development of successful operative repairs and catheter-based corrections performed at an early age, the number of adults with congenital heart disease (CHD) has gradually increased. There are now estimated to be more than a million adults with CHD in the United States and, for the first time in medical history, more adults than children with this problem.

**How is CHD connected to PH?** One of the most common and serious complications that CHD patients develop is pulmonary hypertension (PH). The patients most at risk for this complication are those with so-called “shunt lesions.” These result in blood flowing abnormally from the left side of the heart (the systemic circulation) to the right side of the heart (the pulmonary circulation). Examples of shunts include holes between the upper heart chambers (atrial septal defects) and holes through the lower heart chambers (ventricular septal defects). Usually such shunts are present from birth, but in other cases they have been surgically created as a way to temporarily improve blood flow to the lungs. In either case the presence of excessive blood flow to the pulmonary arteries may trigger the development of increased resistance in the arterial vessels and thus PH. Repairing these lesions early, before they have the chance to cause PH, is important. However, many of these lesions
are not detected until PH has already developed.

Up to 40 percent of CHD patients have structures in the heart that predispose them to the development of PH, and up to 10 percent of patients will actually develop PH. If PH is unrecognized and untreated, it can result in what had traditionally been recognized as “irreversible” damage to the pulmonary vessels and the shunt flow reverses (now traveling from the pulmonary to the systemic circulation). This results in cyanosis (low oxygen delivery to the tissues), a condition that is referred to as Eisenmenger Syndrome. It is now recognized that Eisenmenger Syndrome is not truly irreversible and that medical therapy may improve exercise capacity and possibly even increase survival.

**Why is it important to recognize PH in CHD?** Fortunately the incidence of Eisenmenger Syndrome is now rare in the United States because of improved detection of CHD in children and the availability of simplified and perfected surgical and catheter-based corrections. However, in parts of the world with limited access to healthcare, only 2-15 percent undergo appropriate surgery and up to 30 percent of unrepaired patients develop Eisenmenger Syndrome. This makes CHD-PH a major public health concern.

Even in the absence of Eisenmenger, PH can have a substantial negative health impact. A recent study showed that PH more than doubled the risk of death and heart complications in CHD patients, including heart failure and arrhythmias. In addition, hospitalizations and lengths of stay in the hospital and in the intensive care unit were almost three times higher when PH was present.

The presence of PH can be detected by performing an echocardiogram. Because PH can be due to an intrinsic problem in the lung vessels (pulmonary arterial hypertension) or from backflow from problems on the systemic side of the heart (pulmonary venous hypertension), a heart catheterization is often required to differentiate these two very different problems, as therapy varies greatly depending on the diagnosis.

**What can be done for me?** CHD-PH patients should be closely monitored at regular intervals by physicians experienced in the management of their condition. Although separate guidelines now exist for the treatment of CHD patients and for patients with other forms of PH, treatment strategies are, for the most part, still based more on clinical experience than on study evidence. Eisenmenger patients, in particular, should be managed in centers with experience in the treatment of this syndrome. In general, patients with CHD-PH benefit from a team-based approach to care with input from the CHD specialist and the PH specialist. There are several facets of care that need to be addressed in the CHD-PH patient:

- **Oxygen supplementation:** Many CHD patients, depending on the degree of shunting, have chronic hypoxia (low oxygen in the blood). Some also have varying degrees of lung disease. Oxygen therapy has not been demonstrated to improve exercise tolerance or survival in patients with CHD-PH, but may improve symptoms. Care should be taken not
to over-oxygenate, as improvements in saturation are limited and administration can lead to problems such as nose bleeding when given by nasal cannula.

- **Aerobic exercise** is important to maintain good exercise tolerance, but strenuous exercise generally should be avoided.

- **Anticoagulation:** Although many patients with PH are anticoagulated, this practice in the CHD patient is controversial. Patients with CHD are prone to bleeding – particular lung bleeding or hemoptysis (coughing up blood). When anticoagulation is used, levels of blood thinning should be monitored very carefully to prevent such complications.

- **Diuretics:** Diuretics should be used sparingly in order to avoid dehydration, which can provoke hyperviscosity (abnormal denseness of the blood that can limit blood flow) and can also lead to drops in systemic blood pressure.

- **Treatment of Iron Deficiency:** Despite having higher hematocrits (red blood cell counts) due to low blood oxygen levels (cyanosis), patients with Eisenmenger Syndrome are actually at higher risk for iron deficiency. The latter can increase their risk for hyperviscosity. Phlebotomy (the practice of removing blood to decrease the red blood cell count) should generally be avoided, as this can further worsen iron deficiency. When phlebotomy is absolutely necessary, like when neurological problems result, fluid should always be administered whenever blood is removed.

- **Avoidance of Pregnancy:** CHD-PH is associated with a substantially increased risk for both mother and child during pregnancy. With Eisenmenger Syndrome, there is up to a 50-percent risk of maternal death and a 40-percent risk of spontaneous abortion. Patients should avoid pregnancy and discuss contraceptive options with their doctors.

- **Advanced Medical Therapy:** Randomized controlled trials demonstrating benefits of advanced medical therapy in PH patients have included a small number of patients with CHD-PH (most of whom had undergone a repair of the lesion). There are now reports of use of all three PH drug classes (endothelin antagonists, phosphodiesterase-5 inhibitors and prostanoids) in patients with CHD-PH. In the randomized BREATHE-5 study, bosentan (an endothelin antagonist) improved distance walked and did not appreciably change oxygen saturations in Eisenmenger patients. Recent studies suggest that advanced therapy in CHD-PH may significantly improve quality of life and may even improve survival.

- **Transplantation:** When no other treatments are successful and quality of life is severely impaired, patients with CHD-PH can undergo heart and lung transplantation. Another option is repair of the heart defect along with lung transplantation. The latter, however, is rarely possible or successful. The early death rate after transplantation appears slightly higher than for patients without CHD; but after this early period, most patients experience dramatic improvements in symptoms and quality of life.
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)
- Professional membership sections:
  - PH Clinicians and Researchers (PHCR)—for physicians and doctorate-level researchers
  - PH Professional Network (PHPN)—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.PHAAssociation.org/ConnectOnline)

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Robert Schilz, DO, PhD
*Case Western Reserve University School of Medicine, Cleveland, Ohio*

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**More Information on Congenital Heart Disease**

Adult Congenital Heart Disease Association: [www.ACHAHeart.org](http://www.ACHAHeart.org)

American Heart Association: [www.Heart.org/CongenitalHeartDefects](http://www.Heart.org/CongenitalHeartDefects)