CONDITION

Familial primary pulmonary hypertension

An inherited condition that raises pressure in the lung arteries and strains the heart

WHAT IT IS
Inherited high blood pressure in the lung arteries

BODY SYSTEM
Heart, lungs, and circulation

MAIN SYMPTOMS
Breathlessness, tiredness, chest discomfort, fainting

WHAT CAUSES IT
Inherited gene changes, sometimes unidentified

HOW IT IS DIAGNOSED
Right heart catheterisation after heart and lung tests

MAIN TREATMENT
Pulmonary arterial hypertension medicines and specialist supportive care

CAN IT BE CURED?
No - treatment controls but does not remove it

IS IT AN EMERGENCY?
Sometimes - fainting, chest pain, or severe breathlessness

LONG-TERM OUTLOOK
Treatable but potentially life-shortening, requiring lifelong specialist care

Overview

Familial primary pulmonary hypertension is an older name for heritable pulmonary arterial hypertension , or heritable PAH. It is a rare, long-term condition in which blood pressure becomes too high in the arteries carrying blood from the heart to the lungs.

These arteries become narrowed and less flexible, so the right side of the heart must work harder to pump blood through them. Over time, this strain can weaken the heart.

"Familial" means an inherited gene change may run in a family. Not everyone who inherits a change develops PAH, and current testing does not identify a genetic cause in every affected family.

PAH differs from the common high blood pressure measured with an arm cuff. It is also only one type of pulmonary hypertension. Other types may be caused by left-sided heart

conditions, lung disease, blood clots, or other health problems. Correct classification is important because treatments differ.

Symptoms can resemble asthma, low fitness, anxiety, anaemia, or other heart and lung conditions. Evaluation at a specialist pulmonary hypertension centre is important when PAH is suspected or confirmed.

Symptoms

Symptoms often begin gradually and may initially occur only during exercise, climbing stairs, or walking uphill. They can become noticeable during ordinary activity as the condition progresses.

Common symptoms include:

Symptoms alone cannot show how high the pressure is or identify its cause. Some people have few early symptoms despite significant changes in the lung arteries.

Fainting during activity can mean the heart cannot increase blood flow enough to meet the body's needs. New fainting, severe breathlessness, or chest pain needs prompt medical assessment.

- Shortness of breath during activity
- Unusual tiredness or weakness
- Chest pressure or discomfort
- Dizziness, near-fainting, or fainting
- A racing, pounding, or irregular heartbeat
- Swelling of the ankles, legs, or abdomen
- Reduced ability to exercise
- Bluish or grey lips or fingertips when oxygen levels are low

Causes and risk factors

Familial PAH is associated with inherited changes in genes involved in the growth and function of cells lining the lung arteries. The gene most often involved is BMPR2 , although changes in several other genes can cause or increase susceptibility to PAH.

Inheritance is commonly autosomal dominant, meaning a person with a relevant gene change

may pass it to a child. However, inheriting the change does not mean that PAH will definitely develop. This is called reduced penetrance .

PAH may appear at different ages within the same family. One relative may develop symptoms early, while another may never develop the condition. Women develop heritable PAH more often than men, although anyone can be affected.

Sometimes several relatives have confirmed or suspected PAH but testing does not find a known genetic change. Current tests cannot identify every cause.

Certain medicines, drugs, and health conditions can also cause PAH, but those cases are not automatically familial. Specialist assessment is needed before PAH is classified as heritable.

Diagnosis

Diagnosis involves confirming raised pressure and resistance in the lung arteries while checking for other causes of pulmonary hypertension.

Initial tests may include:

An echocardiogram can estimate pressure and show whether the right side of the heart is enlarged or strained, but it cannot confirm PAH by itself.

Confirmation usually requires right heart catheterisation . A thin tube is passed through a vein into the right side of the heart and lung arteries to measure pressures, blood flow, and resistance. The results help distinguish PAH from pulmonary hypertension caused by left-sided heart problems.

Some people have vasoreactivity testing during catheterisation. This checks the response to a short-acting medicine. Only a small minority have the type of response that makes certain calcium-channel blockers appropriate.

Genetic counselling is recommended when heritable PAH is suspected. A genetics professional can explain the limits of testing, possible effects on relatives, and relevant privacy or insurance considerations.

- An electrocardiogram to assess heart rhythm and electrical activity
- An echocardiogram to examine the heart
- Blood tests for anaemia, liver problems, autoimmune conditions, and other causes
- Breathing tests and chest imaging
- A walking or exercise test

- A ventilation-perfusion scan or other imaging to look for chronic blood clots

Treatment and management

Treatment aims to reduce symptoms, improve activity, slow progression, and protect the right side of the heart. It does not remove the inherited tendency.

Care is usually coordinated through a specialist pulmonary hypertension centre. Treatment depends on symptoms, catheter measurements, heart function, exercise capacity, other conditions, and estimated risk.

PAH-specific medicines work through pathways controlling narrowing and growth in the lung arteries. Categories include:

Medicines may be tablets, inhaled treatments, or continuous infusions under the skin or into a vein. Many people receive a combination of medicines.

Supportive treatment may include diuretics, or water tablets like furosemide or hydrochlorothiazide , for fluid build-up and oxygen when blood oxygen is low. Anticoagulants like warfarin or apixaban are not routine for everyone with heritable PAH because their potential benefits must be weighed against bleeding risks.

Supervised exercise or pulmonary rehabilitation may help when the condition is stable. Activity should be paced. Avoid heavy straining or exercise that causes chest pain, marked breathlessness, or dizziness.

Pregnancy places major stress on the heart and circulation and can be dangerous in PAH. Some PAH medicines can harm a developing baby. People who could become pregnant need specialist advice about contraception, pregnancy planning, and medication safety.

If advanced PAH worsens despite medication, lung or heart-lung transplantation may be considered. Transplantation is a major procedure requiring lifelong follow-up and is not suitable for everyone.

- Endothelin receptor antagonists
- Phosphodiesterase-5 inhibitors
- Soluble guanylate cyclase stimulators
- Prostacyclin-pathway medicines

Complications

The main complication is right-sided heart failure, which occurs when the right side of the heart can no longer pump effectively against high resistance in the lung arteries.

Signs may include worsening breathlessness, swelling, abdominal fullness, rapid weight gain from fluid, and severe tiredness. Other complications include irregular heart rhythms, low oxygen levels, coughing up blood, and fainting.

The condition may also affect sleep, work, relationships, and emotional health. Anxiety or low mood should be addressed as part of PAH care.

Some complications are detected during routine follow-up before severe symptoms develop. Regular assessment helps the specialist team decide whether treatment should change.

Prevention

The inherited tendency to familial PAH cannot currently be prevented. Lifestyle choices do not cause the underlying gene change.

Early detection may be possible in relatives at increased genetic risk. Screening can include symptom review, echocardiography, blood tests, or exercise testing. The schedule depends on family history, genetic findings, age, and specialist assessment.

Genetic counselling can help relatives decide whether to have predictive testing. Testing is usually most informative after a disease-causing change has been identified in a relative with PAH.

People planning a family may wish to discuss reproductive options with genetics and fertility specialists. Counselling should be non-directive.

Avoiding smoking and unregulated stimulant or weight-loss drugs supports heart and lung health, but cannot guarantee that heritable PAH will not develop.

Living with it and what to expect

Familial PAH is usually lifelong. Some people remain stable for years with treatment, while others need treatment to be increased or changed.

Follow-up commonly includes symptom review, physical examination, blood tests, walking tests, echocardiograms, and checks for medicine effects. Right heart catheterisation may be repeated when measurements are needed for major treatment decisions.

Plan activities around your energy level. Gentle, regular movement is often more manageable than occasional intense effort. Rest breaks, workplace adjustments, and practical support can

make daily tasks easier.

Travel may require advance planning, especially for air travel, high altitudes, oxygen, injectable treatment, or medication storage. Carry an up-to-date medicine list and specialist contact details.

Do not stop treatment suddenly without specialist advice. This is particularly important for continuously infused prostacyclin medicines, because an interruption can cause rapid deterioration.

The outlook has improved with newer treatments and combination therapy, but familial PAH can still be life-shortening. Lifelong specialist review helps detect changes early and supports timely decisions about additional treatment or transplant assessment.

When to get medical help

Contact your pulmonary hypertension team if you have a gradual increase in breathlessness, tiredness, swelling, palpitations, or difficulty with activities that were previously manageable. Report persistent medication side effects or problems using an infusion or inhaled treatment.

Seek urgent medical advice for new fainting or near-fainting, new chest discomfort, rapidly increasing swelling, coughing up blood, or a sudden decline in exercise ability.

Call emergency services for:

Do not drive yourself for emergency care.

- Severe breathlessness at rest
- Fainting with slow recovery or injury
- Severe or persistent chest pain
- Blue or grey lips with marked breathing difficulty
- Confusion, collapse, or inability to stay awake
- A sudden interruption of continuous intravenous PAH medicine that cannot be restored immediately

Common questions

Is familial primary pulmonary hypertension the same as pulmonary hypertension?

It is one specific type, now called heritable pulmonary arterial hypertension. Other types result from left-sided heart problems, lung conditions, chronic blood clots, or less common causes.

If my parent has it, will I get it?

Not necessarily. A relevant gene change may be inherited, but not everyone carrying it develops PAH. Genetic counselling can clarify whether testing or screening may be useful.

Can a genetic test rule it out?

No. A positive result can identify an inherited cause and guide family testing, but a negative result does not exclude heritable PAH.

Can I exercise with familial PAH?

Many people can do light or moderate activity when their condition is stable. The safe level is individual. Stop and seek advice if activity causes chest pain, dizziness, fainting, or severe breathlessness.

Can familial PAH be cured by a lung transplant?

Transplantation can replace the lungs containing the narrowed arteries and may be considered for advanced PAH. It involves major surgery, anti-rejection medicines, and lifelong transplant care.

How often do relatives need screening?

There is no single schedule. Screening depends on the identified gene change, family history, age, symptoms, and advice from specialist PAH and genetics teams.

Sources

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