CONDITION

Pulmonary veno-occlusive disease

A rare condition in which narrowed lung veins cause high pressure in the lung circulation.

WHAT IT IS
Blockage of tiny veins carrying blood through the lungs

BODY SYSTEM
Lungs, heart and circulation

MAIN SYMPTOMS
Breathlessness, fatigue, fainting, chest discomfort

WHAT CAUSES IT
Inherited changes, certain exposures, or unknown causes

HOW IT IS DIAGNOSED
Genetic testing or lung tissue; clinical tests strongly support diagnosis

MAIN TREATMENT
Specialist care, oxygen, cautious medicines, lung transplantation

CAN IT BE CURED?
Sometimes - lung transplantation can be definitive

IS IT AN EMERGENCY?
Sometimes - severe breathlessness, fainting, or blue lips

LONG-TERM OUTLOOK
Often progressive; transplantation may improve long-term outlook

Overview

Pulmonary veno-occlusive disease, usually shortened to PVOD, is a rare form of pulmonary hypertension. Pulmonary hypertension means that blood pressure is too high in the blood vessels of the lungs.

In PVOD, the smallest veins that carry blood away from the lungs become narrowed or blocked. The tiny lung blood vessels called capillaries may also be affected. Blood then has difficulty moving through the lungs and back to the left side of the heart.

This pressure makes the right side of the heart work harder to pump blood. It may also cause fluid to leak into lung tissue, known as pulmonary oedema. Over time, PVOD can lead to low oxygen levels and right-sided heart failure.

PVOD can look very similar to pulmonary arterial hypertension, or PAH. The distinction matters

because medicines that widen lung arteries can sometimes trigger serious fluid build-up in people with PVOD. Assessment and treatment therefore need to be led by a pulmonary hypertension specialist centre.

Symptoms

Symptoms often develop gradually. Early changes may be noticed only during exercise, climbing stairs, or walking uphill. As the condition progresses, ordinary activities may become more difficult.

Common symptoms include:

Low oxygen may cause the lips or fingertips to look blue or grey, although this can be harder to see on darker skin. Some people develop rounded or enlarged fingertips, called clubbing.

When the right side of the heart is struggling, swelling may appear in the ankles, legs, or abdomen. Rapid weight gain can occur because the body is retaining fluid.

Symptoms alone cannot show whether someone has PVOD. Several heart and lung conditions can cause similar problems.

- Shortness of breath, especially during activity
- Unusual tiredness or weakness
- Dizziness or fainting
- Chest pressure or discomfort
- A racing or pounding heartbeat
- A dry cough
- Reduced ability to exercise

Causes and risk factors

PVOD may be inherited, linked to an exposure or another health condition, or have no identifiable cause.

Inherited PVOD is associated with harmful changes in both copies of a gene called EIF2AK4 . Genes are instructions carried in your DNA. This form follows an autosomal recessive pattern, meaning a person usually inherits one altered copy from each parent. The parents are often healthy carriers.

Having one altered copy does not usually mean that you will develop inherited PVOD. A

genetics professional can explain what a result may mean for you and your relatives.

Many people with PVOD do not have an identified inherited cause. In these cases, the condition is described as sporadic.

PVOD-like changes have also been reported in association with:

These associations do not mean that everyone with one of these exposures will develop PVOD. In many cases, it is not possible to identify why the condition occurred.

- Certain chemotherapy medicines, particularly mitomycin
- Bone marrow transplantation
- Exposure to some organic solvents
- Radiation involving the chest
- Connective tissue conditions, especially systemic sclerosis
- Tobacco exposure

Diagnosis

PVOD can be difficult to diagnose because its symptoms and heart test results often resemble PAH. Evaluation is usually coordinated by a specialist pulmonary hypertension team.

An echocardiogram uses sound waves to examine the heart. It can show strain on the right side of the heart and estimate pressure in the lung circulation, but it cannot confirm PVOD.

A CT scan of the chest may show a combination of clues. These include thickened lines between sections of lung, hazy areas called ground-glass opacities, enlarged lymph nodes in the chest, and a widened pulmonary artery. No single CT finding proves the diagnosis.

Lung function tests measure how well air and gases move through the lungs. People with PVOD often have a particularly low diffusing capacity, or DLCO , which means oxygen does not pass efficiently from the lungs into the blood. Blood tests or a finger sensor may also show a low oxygen level.

Right-heart catheterisation is used to confirm pulmonary hypertension. A thin tube is passed through a vein into the right side of the heart and lung arteries to measure pressures directly. PVOD usually produces a pattern called precapillary pulmonary hypertension. The pressure measured on the left side of the lung circulation may appear normal even though disease is present in the smallest veins.

Genetic testing can identify harmful changes in both copies of EIF2AK4 . In the right clinical setting, this can establish a heritable diagnosis without a lung biopsy.

Examining lung tissue can confirm PVOD, but taking a surgical lung biopsy is usually avoided. It may cause severe bleeding or a dangerous decline in breathing. Tissue confirmation is therefore most often made after lung transplantation or at post-mortem examination.

Treatment and management

There is no medicine that reliably reverses the blocked lung veins. Management focuses on supporting breathing and heart function, controlling symptoms, and assessing whether lung transplantation is suitable.

Because PVOD is uncommon and reacts differently from many other forms of pulmonary hypertension, treatment should be planned through an expert centre.

Oxygen may be used when blood oxygen levels are low. Some people need it during activity or sleep, while others need it more continuously.

Diuretics like furosemide or hydrochlorothiazide , sometimes called water tablets, may help the body remove excess fluid. They can reduce leg swelling and ease the workload on the heart. Blood pressure, kidney function, body weight, and blood salts may need monitoring during treatment.

Medicines used for PAH widen blood vessels in the lungs. They may improve symptoms or circulation in carefully selected people with PVOD, sometimes as support while awaiting transplantation. However, they can also cause pulmonary oedema because more blood enters lungs whose small veins cannot drain it properly. Specialists may therefore introduce treatment cautiously and monitor closely for worsening breathlessness or fluid in the lungs.

Lung transplantation is the only treatment that can replace the affected lung circulation. Referral is often considered early because PVOD may progress and transplant assessment takes time. Eligibility is based on several factors, including general health, heart function, other medical conditions, and the likely balance of risks and benefits.

Supportive care may include supervised rehabilitation, vaccinations to reduce respiratory infection risk, nutrition support, and help with the emotional effects of living with a serious condition. Any exercise plan needs to reflect oxygen levels, symptoms, and specialist advice.

Complications

PVOD can place increasing strain on the right side of the heart. This may lead to right-sided heart failure, in which the heart cannot move blood forward effectively.

Other possible complications include:

Pulmonary oedema may occur as part of the condition or after starting a medicine that widens lung arteries. New breathing difficulty after a treatment change needs prompt medical assessment.

- Persistently low blood oxygen
- Pulmonary oedema, meaning fluid within the lungs
- Fainting caused by reduced blood flow during exertion
- Abnormal heart rhythms
- Coughing up blood
- Reduced kidney or liver function from advanced heart failure

Prevention

Inherited PVOD cannot currently be prevented. Genetic testing and counselling cannot remove the condition, but they may clarify risks for relatives and support family-planning decisions.

There is no proven way to prevent sporadic PVOD when the cause is unknown. Avoiding tobacco smoke and following workplace protections around solvents can reduce other lung and heart risks, but these steps have not been shown to prevent every case of PVOD.

If a medicine or cancer treatment has been linked with PVOD, do not stop it on your own. The treating team can balance its benefits against the risk and investigate new breathing symptoms.

Living with it and what to expect

PVOD is generally progressive, meaning changes in the lung vessels may continue over months or years. The speed is not the same for everyone, and tests at one point cannot predict an exact course.

You may need regular reviews involving walking tests, oxygen measurements, blood tests, heart scans, and discussions about symptoms. The team may also track weight and swelling because these can show fluid retention. Follow-up is often more frequent after a medicine is started or adjusted.

Daily activities may need to be paced. Resting between tasks and avoiding sudden heavy

exertion can help manage breathlessness. Some people can continue working with adjustments, while others need reduced hours or changes in duties. Oxygen equipment and travel planning may require practical preparation.

Pregnancy places major extra strain on the heart and lung circulation and carries serious risk in pulmonary hypertension. Anyone who could become pregnant should have access to specialist advice about contraception and pregnancy planning.

Waiting for transplant assessment or living with uncertainty can affect sleep, relationships, and mood. Psychological support, social work services, and patient groups may help with practical and emotional concerns.

Transplantation can substantially change the outlook for eligible people, but it involves major surgery and lifelong follow-up. Anti-rejection medicines are needed afterward. Not everyone can receive a transplant, and results cannot be guaranteed.

When to get medical help

Call emergency services if you have:

Seek urgent same-day advice from your specialist team for a clear increase in breathlessness, new swelling, rapid weight gain, lower oxygen readings than usual, or new symptoms after starting or increasing a pulmonary hypertension medicine.

Routine follow-up remains important when symptoms are stable. Bring changes in exercise ability, dizziness, oxygen use, swelling, side effects, and missed doses to planned appointments rather than waiting for symptoms to become severe.

- Severe or rapidly worsening breathlessness, especially at rest
- Fainting or difficulty waking
- Blue or grey lips, face, or tongue
- New severe chest pain
- Confusion or extreme weakness
- Coughing up more than a small streak of blood
- Breathlessness with frothy sputum, which may indicate pulmonary oedema

Common questions

Is pulmonary veno-occlusive disease the same as pulmonary arterial hypertension?

No. Both cause high pressure in the lung circulation, but they affect different parts of the blood vessel system. PAH mainly affects small arteries entering the lungs. PVOD mainly affects small veins leaving the lung capillaries. Standard PAH medicines therefore need extra caution in PVOD.

Can PVOD be cured?

Lung transplantation can replace the affected lungs and is considered the only potentially definitive treatment. Medicines and oxygen may support the heart and lungs, but they do not remove the underlying vein blockage.

Does PVOD run in families?

Some cases do. Harmful changes in both copies of the EIF2AK4 gene can cause inherited PVOD. Many people have no known family history, and not every case has an identified genetic cause.

Why is a lung biopsy usually avoided?

A biopsy can confirm the pattern of blocked veins, but removing lung tissue can cause major bleeding or breathing problems in someone with severe pulmonary hypertension. Specialists usually rely on the combined results of scans, heart catheterisation, lung tests, and genetic testing.

Can I exercise with PVOD?

Some gentle or supervised activity may be helpful, but strenuous exercise can cause severe breathlessness, dizziness, or fainting. A specialist team can help set safe limits based on symptoms and oxygen levels. Stop activity and seek advice if you develop chest discomfort, marked dizziness, or unusual breathlessness.

Why can pulmonary hypertension medicine make PVOD worse?

These medicines may increase blood flow into the lungs. If narrowed veins cannot carry that blood away, pressure can build behind the blockage and fluid may leak into lung tissue. This is why such treatment requires cautious specialist monitoring.

Sources

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