CONDITION

Familial emphysema

Familial emphysema is an inherited tendency to develop emphysema, often linked to alpha-1 antitrypsin deficiency.

WHAT IT IS
Inherited susceptibility to emphysema, often from alpha-1 antitrypsin deficiency

BODY SYSTEM
Lungs and breathing

MAIN SYMPTOMS
Breathlessness, cough, wheeze, reduced exercise tolerance

WHAT CAUSES IT
Inherited gene changes, especially SERPINA1 variants

HOW IT IS DIAGNOSED
Lung tests plus alpha-1 blood and genetic testing

MAIN TREATMENT
Lung protection, inhalers, rehabilitation, selected replacement therapy

CAN IT BE CURED?
No - damaged air sacs cannot regrow

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

LONG-TERM OUTLOOK
Often manageable; progression reflects damage and ongoing exposures

Overview

Familial emphysema means emphysema occurs more often than expected within a family. It is not one precisely defined diagnosis. The best-known inherited cause is alpha-1 antitrypsin deficiency , also called AAT deficiency or AATD.

Emphysema is a form of chronic obstructive pulmonary disease, or COPD. It damages the walls between the tiny air sacs in the lungs, reducing their natural stretch and making it harder to move air out and exchange oxygen.

Alpha-1 antitrypsin is a protective protein made mainly in the liver. Certain inherited changes in the SERPINA1 gene can lower the amount of this protein or affect how it works. This leaves lung tissue more vulnerable to damage, especially from tobacco smoke, dust, fumes, and repeated inflammation.

Having a relative with emphysema does not mean you will develop it. Families may share genes and environmental exposures, and some people with inherited susceptibility never develop significant breathing problems. Identifying an inherited cause may affect testing, treatment, liver monitoring, and whether adult relatives consider testing.

Symptoms

Emphysema usually develops gradually, and early damage may cause no clear symptoms.

Possible symptoms include:

Symptoms cannot show whether emphysema is inherited. Asthma, heart conditions, infections, and other problems can cause similar symptoms.

AAT deficiency may also cause liver problems because abnormal protein can collect in liver cells. Possible signs include yellow skin or eyes, abdominal swelling, itching, or dark urine. These require medical assessment. Rarely, AAT deficiency is associated with panniculitis , a painful inflammation of fat beneath the skin.

- Shortness of breath during activity
- A long-lasting cough, with or without mucus
- Wheezing or chest tightness
- Reduced stamina
- Frequent chest infections or breathing flare-ups
- Unplanned weight loss in advanced emphysema

Causes and risk factors

Familial emphysema is most clearly linked to inherited variants in the SERPINA1 gene. You receive one copy from each biological parent. Different combinations of variants produce different protein levels and risks.

People with two variants that greatly reduce alpha-1 antitrypsin generally have the highest risk of AAT-related lung damage. Carrying one altered copy may have a smaller effect, but smoking and other exposures can increase the risk. Genetic results must be considered alongside protein levels, lung tests, symptoms, and exposure history.

Tobacco smoke is a major risk factor. It increases inflammation and can disable some available alpha-1 antitrypsin, causing earlier and faster lung damage.

Other risk factors include:

Not every family cluster is caused by AAT deficiency. Emphysema may result from several genes combined with shared environments. If AAT testing is normal, there may be no single genetic test that explains the family history.

- Second-hand smoke
- Workplace dust, chemical fumes, or vapours
- Biomass smoke, such as smoke from indoor fires
- Repeated respiratory infections
- Asthma or other ongoing airway inflammation

Diagnosis

A healthcare professional will ask about your breathing, smoking and work exposures, and relatives with emphysema, COPD, unexplained liver problems, or known AAT deficiency.

Spirometry measures how much air you can blow out and how quickly. It is usually repeated after you inhale medicine that opens the airways. Persistent difficulty blowing air out supports a COPD diagnosis, although spirometry cannot identify the genetic cause.

A chest CT scan can show damaged air sacs and their location. AAT-related emphysema often affects the lower lungs, although not in everyone. A chest X-ray can miss early changes.

Testing for AAT deficiency usually involves:

A protein level alone may not be enough because alpha-1 antitrypsin can rise during infection or inflammation, and levels can overlap between genetic types. Genetic or protein testing helps confirm the result.

Other assessments may include oxygen measurements, full lung-function tests, liver blood tests, or liver imaging. Genetic counselling can explain what a confirmed result may mean for you and your relatives.

- A blood test measuring alpha-1 antitrypsin
- Genetic testing for SERPINA1 variants
- Protein typing, or phenotyping

Treatment and management

Emphysema damage cannot currently be reversed, but treatment can reduce symptoms, protect remaining lung function, and limit flare-ups.

Avoiding tobacco smoke is especially important. If you smoke, counselling and stop-smoking medicines can help. Reducing workplace dust or fumes may require ventilation, protective equipment, or occupational health advice.

Inhaled medicines may open the airways and ease breathing. Some people also need inhaled medicines that reduce airway inflammation. These treatments manage COPD symptoms but do not correct the inherited deficiency.

Pulmonary rehabilitation combines supervised exercise, breathing support, and education. It can improve fitness and help you manage breathlessness, although it cannot rebuild damaged air sacs.

Other management may include:

Selected adults with severe AAT deficiency and emphysema may be offered augmentation therapy , involving regular infusions of purified alpha-1 antitrypsin. It may slow loss of lung tissue in some people, but it does not restore damaged lung or reliably remove existing symptoms. Eligibility and availability vary.

For advanced emphysema, specialists may consider procedures to reduce overinflated lung areas or lung transplantation. These options require detailed assessment and suit only some people.

- Recommended vaccinations against respiratory infections
- A written plan for COPD flare-ups
- Oxygen when tests show persistently low oxygen levels
- Nutrition support for weight loss or difficulty eating
- Treatment and monitoring of liver problems

Complications

Progressive emphysema can make everyday activities increasingly difficult. Periods of suddenly worse cough, mucus, or breathlessness are called exacerbations or flare-ups. Infections and air pollution are common triggers.

Possible complications include:

AAT deficiency can also cause liver scarring, cirrhosis, or liver cancer. The severity of lung symptoms does not reliably predict whether liver problems are present.

- Low blood oxygen
- High pressure in the lung blood vessels
- Strain on the right side of the heart
- Respiratory failure
- A collapsed lung caused by leaking air
- Anxiety, low mood, or reduced confidence with activity

Prevention

You cannot prevent inheriting a SERPINA1 variant or another genetic susceptibility, but you may be able to delay or prevent some lung damage.

The most effective protective step is not smoking. Avoiding second-hand smoke, dust, chemical fumes, and poorly ventilated combustion smoke can also reduce lung injury.

Assessment of persistent breathing symptoms may identify airflow obstruction before it becomes advanced. Recommended vaccinations can lower the risk of infections that trigger flare-ups.

When AAT deficiency is confirmed, biological relatives can be offered information about testing. Testing is a personal choice, and genetic counselling can help families understand inheritance, privacy, and the possible effects of knowing a result.

Living with it and what to expect

The course varies. Some people remain stable for years, while others gradually lose lung function. Outlook depends on existing damage, genetic type, smoking and other exposures, infections, and general health.

Stopping smoking can slow further loss of lung function even after emphysema develops. Exercise and pulmonary rehabilitation can improve daily function without changing the damaged areas.

Regular follow-up commonly includes reviewing symptoms, flare-ups, inhaler use, activity, and oxygen needs. Lung-function tests may be repeated to track changes. People with confirmed AAT deficiency may also need liver monitoring.

Breathlessness can lead to inactivity, which weakens muscles and makes exertion harder. A planned exercise programme, pacing, rest breaks, and breathing techniques can help interrupt

this cycle.

Many people continue working, travelling, exercising, and caring for family. Adjustments may become necessary if symptoms progress. If oxygen is prescribed, portable systems may support activity when medically appropriate.

When to get medical help

Arrange a routine appointment if you have persistent breathlessness, cough, wheezing, or reduced exercise tolerance. Assessment is also reasonable if emphysema, early-onset COPD, AAT deficiency, or unexplained liver problems occur in your biological family.

Contact your usual healthcare service promptly if your breathing becomes worse than normal, your mucus increases or changes colour, you develop a fever, or your usual treatment is not helping. These may indicate a flare-up or infection.

Seek emergency help for:

Do not drive yourself for emergency care if you are severely short of breath or confused.

- Severe breathlessness at rest or difficulty speaking
- Blue or grey lips, skin, or fingertips
- New confusion, extreme drowsiness, or fainting
- Sudden chest pain, especially with abrupt breathlessness
- Coughing up more than a small streak of blood

Common questions

Is familial emphysema the same as alpha-1 antitrypsin deficiency?

Not always. AAT deficiency is the best-established inherited cause. Emphysema can also cluster in families because of multiple genes combined with shared smoking, workplace, or household exposures.

Can I develop familial emphysema if I have never smoked?

Yes. Severe AAT deficiency can cause emphysema in people who have never smoked. Smoking greatly increases the risk and often causes earlier or faster damage. Other inhaled pollutants can also contribute.

What age does inherited emphysema start?

There is no single age. Symptoms may appear in early or middle adulthood, but some people remain well until later life. Smoking, other exposures, genetic type, and the amount of lung damage affect when symptoms begin.

Should my family be tested?

When AAT deficiency is confirmed, biological relatives may be offered testing because they could carry the same variant. They should receive clear information about what results can and cannot predict. A family history without confirmed AAT deficiency does not automatically identify a testable gene.

Can damaged lungs heal after I stop smoking?

Stopping smoking reduces ongoing injury and can slow further decline, but destroyed air-sac walls do not regrow. Cough, airway irritation, fitness, and breathing control may still improve.

Is familial emphysema contagious?

No. Emphysema and AAT deficiency cannot spread between people. Respiratory infections that trigger flare-ups may be contagious, but the underlying emphysema is not.

Sources

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