CONDITION

Familial dilated cardiomyopathy

An inherited condition in which the heart's main pumping chamber enlarges and becomes weaker.

WHAT IT IS
Inherited enlargement and weakening of the heart muscle

BODY SYSTEM
Heart and circulation

MAIN SYMPTOMS
Breathlessness, tiredness, swelling, irregular heartbeats

WHAT CAUSES IT
Inherited gene changes

HOW IT IS DIAGNOSED
Heart imaging, family assessment, sometimes genetic testing

MAIN TREATMENT
Heart-failure medicines, monitoring, and sometimes implanted devices

CAN IT BE CURED?
No - treatment can protect heart function

IS IT AN EMERGENCY?
Sometimes - severe breathing trouble or fainting

LONG-TERM OUTLOOK
Often manageable, with ongoing progression and rhythm risks

Overview

Familial dilated cardiomyopathy, sometimes called familial congestive cardiomyopathy or FDC, is an inherited heart muscle condition. "Dilated" means that a heart chamber has become enlarged. "Cardiomyopathy" means a condition affecting the heart muscle.

The left ventricle, the heart's main pumping chamber, is usually affected most. As it stretches and weakens, it may not pump blood as effectively. This can lead to heart failure, meaning the heart cannot meet the body's needs as well as it should. It can also cause abnormal heart rhythms.

"Familial" means the condition occurs within a family. Some people develop symptoms early, while others remain well for many years. Even relatives with the same genetic change may be affected differently.

Early recognition matters because monitoring and treatment can protect heart function, control symptoms, and identify dangerous heart rhythms. It may also allow relatives at risk to be assessed before symptoms develop.

Symptoms

Familial dilated cardiomyopathy may cause no symptoms at first. It may be found during family screening or after an abnormal heart test.

When symptoms occur, they can include:

Symptoms often result from reduced pumping strength or fluid building up in the lungs and body. Palpitations, dizziness, and fainting may be caused by an abnormal heart rhythm.

These symptoms have many possible causes. Having one does not mean that you have familial dilated cardiomyopathy.

- Shortness of breath during activity or while lying flat
- Unusual tiredness or reduced ability to exercise
- Swelling of the ankles, legs, or abdomen
- A fast, pounding, or irregular heartbeat
- Dizziness, light-headedness, or fainting
- Persistent coughing, especially at night
- Chest discomfort

Causes and risk factors

Familial dilated cardiomyopathy is usually caused by an inherited change in a gene involved in heart muscle structure or function. Many different genes can be involved. Changes in genes such as TTN and LMNA are among those linked to the condition, but genetic testing does not find a cause in every family.

Inheritance is often autosomal dominant. This means a person with the genetic change has a 1 in 2 chance of passing that change to each child. Inheriting a change does not always mean someone will develop heart problems. This is called reduced penetrance.

Symptoms and severity can also differ between relatives. This is called variable expression. One relative may have mild changes, while another develops heart failure or rhythm problems.

Other factors can add strain to a genetically vulnerable heart. These may include pregnancy,

heavy alcohol use, some infections, certain cancer treatments, and poorly controlled high blood pressure. These factors do not explain every case, and exposure does not mean the condition will develop.

Sometimes several relatives have dilated cardiomyopathy even though genetic testing has not identified a specific change. A detailed family history remains important in this situation.

Diagnosis

Dilated cardiomyopathy is diagnosed by showing that the heart is enlarged and pumps less effectively, while checking for other explanations. Establishing that it is familial requires information about relatives and, when informative, genetic testing.

Assessment may include:

A genetic test may identify a pathogenic variant, meaning a gene change known to cause the condition. A negative result does not rule out a familial cause because not every responsible gene change is currently detectable or understood.

A variant of uncertain significance is a gene change whose meaning is not yet clear. By itself, it should not be treated as confirmation that a relative has the condition.

If a causative variant is found, close relatives may be offered testing for that specific variant. When no variant is found, relatives may still need periodic ECGs and heart imaging based on specialist advice.

- An electrocardiogram, or ECG, to record the heart's electrical activity
- An echocardiogram, which uses ultrasound to assess heart size and pumping
- Cardiac MRI for more detailed images and signs of heart muscle scarring
- Blood tests for conditions that can affect the heart
- Heart-rhythm monitoring over one or more days
- Tests for narrowed heart arteries when this is a possible cause
- A medical history covering symptoms, pregnancy, alcohol, medicines, and treatments
- A family history of cardiomyopathy, heart failure, rhythm problems, or sudden death

Treatment and management

Treatment aims to support the heart, reduce symptoms, prevent complications, and slow

worsening. The plan is based on pumping function, symptoms, heart rhythm, genetic findings, and other health conditions.

People with reduced heart pumping function may be offered several types of heart-failure medicine. These can include beta blockers like metoprolol or propranolol , medicines acting on the renin-angiotensin system, mineralocorticoid receptor antagonists, and SGLT2 inhibitors. Diuretics like furosemide or hydrochlorothiazide , sometimes called water tablets, may help when fluid causes swelling or breathlessness.

Medicines are usually introduced and adjusted with blood pressure, kidney function, symptoms, and blood tests in mind. Do not stop heart medicines because you feel better unless the prescribing team advises it. Heart function can worsen again after treatment is withdrawn.

An implantable cardioverter-defibrillator, or ICD, can detect and treat certain dangerous heart rhythms. It may be considered when pumping function remains significantly reduced or when genetic and rhythm findings indicate a higher risk. A cardiac resynchronisation therapy device can help selected people whose heart chambers are not beating in a coordinated way.

Other parts of management may include:

Very intense exercise may increase rhythm risk in some genetic forms. This does not mean everyone should avoid physical activity. The safest type and level of exercise should be based on heart function, rhythm history, symptoms, and the gene involved.

For severe heart failure that does not respond to standard treatment, a mechanical heart pump or heart transplant may be considered.

- Treatment for atrial fibrillation or other abnormal rhythms
- Anticoagulant medicine when there is a specific risk of blood clots
- Cardiac rehabilitation and an individually suitable activity plan
- Advice about alcohol, salt, fluid intake, smoking, and weight
- Pregnancy planning with cardiology and maternity specialists
- Genetic counselling for you and your family

Complications

Possible complications include:

The chance of these problems is not the same for everyone. Regular follow-up helps identify changes before they cause major symptoms.

- Heart failure with fluid in the lungs or legs
- Atrial fibrillation or other abnormal heart rhythms
- Life-threatening rhythms arising from the ventricles
- Blood clots, stroke, or clots travelling elsewhere
- Heart valve leakage as the heart enlarges
- Progressive loss of pumping function

Prevention

The inherited tendency to familial dilated cardiomyopathy cannot currently be prevented. A person can carry a causative gene change despite having a healthy lifestyle.

Early family assessment may detect heart changes before symptoms appear. Treatment can then begin when appropriate. Avoiding heavy alcohol use, not smoking, and managing blood pressure may reduce additional strain on the heart, but these steps cannot remove the genetic risk.

Genetic counselling can explain inheritance, testing choices, and reproductive options. It does not tell with certainty whether or when a person carrying a gene change will develop heart problems.

Living with it and what to expect

The course is different between individuals and families. Some people remain stable for years with monitoring and treatment. Others experience a gradual decline in pumping function, repeated fluid retention, or rhythm problems. Certain genetic forms carry a higher rhythm risk even before the heart becomes severely enlarged.

Treatment can sometimes produce "reverse remodelling," meaning the heart becomes smaller and pumps more strongly. This is an improvement rather than proof that the inherited tendency has gone away. Ongoing follow-up and treatment may still be needed.

Follow-up commonly includes discussion of symptoms, an ECG, heart imaging, blood tests, and sometimes rhythm monitoring. The schedule is based on your findings and can change over time.

Many people continue working, exercising at a suitable level, travelling, and having a family. Plans may need adjustment during symptom flare-ups or after a device procedure. Pregnancy

can place extra demands on the heart, so planning before pregnancy is important.

Family information may also change. A gene variant can be reclassified as research develops, or a newly diagnosed relative may clarify the family pattern. Keeping the cardiology or genetics service updated about major family events can be useful.

When to get medical help

Arrange a routine medical review if you develop gradually increasing breathlessness, ankle swelling, tiredness, palpitations, or reduced exercise ability. Also seek routine assessment if a close relative has been diagnosed with dilated cardiomyopathy or an associated genetic variant, even if you feel well.

Contact your heart team promptly if established symptoms noticeably worsen, your weight rises quickly with swelling, you become short of breath at rest, or you receive an alert or shock from an implanted heart device.

Call emergency services for:

Do not drive yourself to emergency care if you are faint, severely breathless, or having possible stroke or heart attack symptoms.

- Severe breathing difficulty, especially at rest
- Fainting or collapse
- Chest pain that is severe, persistent, or accompanied by sweating or nausea
- A sustained rapid heartbeat with dizziness, weakness, or breathlessness
- Signs of stroke, such as facial drooping, arm weakness, or speech difficulty

Common questions

Is familial dilated cardiomyopathy the same as heart failure?

No. It is a heart muscle condition that can cause heart failure, but some people have mild heart changes without heart-failure symptoms. Heart failure describes how well the heart is meeting the body's needs, not a separate diagnosis that everyone with cardiomyopathy automatically has.

Can familial dilated cardiomyopathy skip a generation?

It can appear to. A person may inherit a causative variant but never develop clear symptoms, or changes may not appear until later in life. That person can still pass the variant to a child.

Will my children inherit it?

For many autosomal dominant forms, each child of a person carrying the variant has a 1 in 2 chance of inheriting it. Other inheritance patterns also occur. Inheriting a variant does not predict the age of onset or severity. Genetic counselling can explain the pattern in your family.

Can my heart return to normal?

Heart size and pumping strength can improve substantially with treatment, and some measurements may return to the normal range. This does not necessarily mean the underlying tendency has been cured. Continued treatment and monitoring may still be advised.

Can I exercise with familial dilated cardiomyopathy?

Many people can remain active, but the safest intensity is individual. Symptoms, pumping function, abnormal rhythms, implanted devices, and the gene involved all matter. A cardiology team or cardiac rehabilitation service can help define a suitable plan.

Can I still have it if my genetic test is negative?

Yes. Current testing does not identify every genetic cause. Diagnosis and family screening may still be based on heart imaging, electrical tests, personal history, and the pattern of heart problems among relatives.

Sources

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