CONDITION

EDMD

EDMD is an inherited condition that causes muscle tightening, weakness, and potentially serious heart rhythm problems.

WHAT IT IS
Inherited muscular dystrophy affecting muscles, joints, and heart

BODY SYSTEM
Muscles, joints, heart and circulation

MAIN SYMPTOMS
Joint tightening, muscle weakness, abnormal heartbeat, fainting

WHAT CAUSES IT
Inherited or new genetic changes

HOW IT IS DIAGNOSED
Genetic testing, muscle assessment, and heart tests

MAIN TREATMENT
Therapy, mobility support, and heart rhythm management

CAN IT BE CURED?
No, but complications can be managed

IS IT AN EMERGENCY?
Sometimes, especially with fainting or severe breathlessness

LONG-TERM OUTLOOK
Often slowly progressive with lifelong heart monitoring

Overview

EDMD stands for Emery-Dreifuss muscular dystrophy. It is an inherited condition affecting skeletal muscles, joints, and the heart.

EDMD usually has three main features:

Joint tightening is called a contracture . It often affects the elbows, ankles, and back of the neck and may appear before obvious weakness.

Muscle weakness may begin in the upper arms and lower legs before affecting the shoulders, hips, and other areas.

Heart involvement is especially important. The heart may beat too slowly, irregularly, or less effectively. This can develop even when muscle symptoms are mild, so lifelong heart checks

are needed even for people who feel well.

EDMD often progresses slowly, and many people remain active and independent for years.

- Tight joints that cannot move through their full range
- Slowly progressive muscle weakness
- Problems with the heart's rhythm or pumping ability

Symptoms

Symptoms may begin in childhood or adolescence, although some people are diagnosed later. Their timing and severity can differ even among relatives with the same genetic change.

Early contractures commonly affect:

Tight Achilles tendons can make it difficult to place the heels flat on the floor. Elbow contractures may prevent the arms from straightening fully, while neck or spinal stiffness can limit movement and cause a rigid posture.

Weakness often affects the upper arms and lower legs first. This may cause difficulty running, climbing stairs, lifting the arms, or walking on the heels. Shoulder and hip weakness may become more noticeable later.

Heart problems may initially cause no symptoms. When symptoms occur, they can include:

Some people develop cardiomyopathy , a disease of the heart muscle that can reduce its ability to pump. Others mainly have problems with the electrical system controlling the heartbeat.

Breathing problems are less common than in some muscular dystrophies, but breathing muscle weakness, spinal stiffness, or disturbed breathing during sleep can occur.

- The Achilles tendons at the backs of the ankles
- The elbows
- The neck and spine
- A racing, fluttering, or skipping heartbeat
- Unusual tiredness or reduced exercise tolerance
- Dizziness or fainting
- Shortness of breath
- Swelling in the legs or abdomen

Causes and risk factors

EDMD is caused by a harmful change in a gene involved in muscle and heart function. Many associated proteins support the nuclear envelope, the structure surrounding a cell's control centre.

Two well-established EDMD genes are:

Changes in other genes can produce an EDMD-like pattern. Genetic testing matters because the gene may affect inheritance, heart risks, and testing recommendations for relatives.

EMD-related EDMD is usually X-linked. It most often causes muscle symptoms in males, but females carrying the genetic change can develop heart rhythm problems and sometimes muscle symptoms.

LMNA-related EDMD is often autosomal dominant, meaning one changed gene copy can cause the condition. Less commonly, EDMD is autosomal recessive, requiring changes in both copies.

A person may also have a new, or de novo , genetic change that was not found in either parent. No known family history therefore does not rule out EDMD.

EDMD is not caused by exercise, diet, infection, or something a parent did during pregnancy.

- EMD , which provides instructions for a protein called emerin
- LMNA , which provides instructions for lamin A and lamin C

Diagnosis

Diagnosis involves a medical and family history and an examination of muscle strength, joint movement, posture, and walking. The pattern may suggest EDMD, but symptoms alone cannot identify the gene involved.

Genetic testing can confirm the diagnosis in many people. It may examine a panel of genes linked to muscular dystrophy and inherited heart conditions. Broader testing may be needed if the first test does not find a cause.

Blood tests may measure creatine kinase (CK), an enzyme released when muscle is damaged. CK may be raised in EDMD, but the result is not specific and can be only mildly abnormal.

Other tests may include electromyography or muscle MRI. Muscle biopsy is now less commonly needed but may be considered if genetic results are unclear.

Heart assessment is required at diagnosis and commonly includes:

Close relatives may be offered genetic counselling, genetic testing, or heart screening,

depending on the result.

- An electrocardiogram, or ECG
- Longer rhythm monitoring, such as a Holter or patch monitor
- An echocardiogram
- Sometimes cardiac MRI to assess the heart muscle and scarring

Treatment and management

There is no current cure that reverses EDMD. Treatment aims to preserve movement and independence, manage symptoms, and reduce heart-related risks.

Physiotherapy can help maintain joint movement and support safe activity. A therapist may recommend gentle stretching and suitable exercise. Forceful stretching can injure muscles or joints, so plans should be individually adapted.

Braces or splints may support the ankles and improve walking. Occupational therapy can help with daily activities, school, work, and equipment. Surgery may be considered for severe contractures, although tightening can return and rehabilitation is important.

Moderate, low-impact activity is generally encouraged within personal limits. Repeated exercise to exhaustion or heavy strength training may worsen pain or muscle damage in some people.

Heart care is central to treatment. A pacemaker may be used if the heartbeat is dangerously slow or electrical signals are blocked. An implantable cardioverter-defibrillator (ICD) can treat certain dangerous fast rhythms. Device choice depends on detailed heart assessment and genetic findings.

Medicines may be used for cardiomyopathy or heart failure. Some people with advanced heart disease may be assessed for heart transplantation.

Breathing tests or sleep studies may be recommended for morning headaches, poor sleep, daytime sleepiness, or breathlessness. Assisted ventilation, often through a mask at night, can support breathing when needed.

Complications

The most serious complications involve the heart. Electrical conduction disease can make the heart beat too slowly or pause, while other rhythm problems cause a very fast heartbeat.

These problems may lead to dizziness, fainting, blood clots, stroke, or sudden cardiac arrest.

Abnormal rhythms in the heart's upper chambers can allow blood clots to form. Treatment may include medicine to reduce stroke risk.

Cardiomyopathy can lead to heart failure, causing tiredness, breathlessness, swelling, or reduced exercise tolerance.

Progressive contractures and weakness can affect walking, reaching, posture, and comfort. They may lead to falls or the need for mobility equipment. Some people eventually use a wheelchair, while others remain able to walk.

Spinal stiffness and breathing muscle weakness can reduce breathing capacity. Reduced mobility can also affect bone strength.

Prevention

EDMD itself cannot usually be prevented because it results from a genetic change. Nothing a person with EDMD or their parents did caused it.

Regular care can prevent or identify some complications early. Heart rhythm monitoring is especially important because serious problems may develop before symptoms appear. Stretching, mobility support, and prompt contracture management may help preserve function.

Genetic counselling can explain inheritance, testing for relatives, and reproductive options. These are personal choices, and counselling provides information rather than directing decisions.

Living with it and what to expect

EDMD is lifelong. Weakness and contractures often progress slowly, but the pace varies. The gene involved, the specific genetic change, and heart involvement all affect the course.

Many people continue with education, work, relationships, exercise, and family life. Braces, accessible transport, flexible working, or mobility equipment may reduce fatigue and support independence. Using equipment does not mean all walking or activity must stop.

Contractures do not always match the degree of weakness. Someone may have tight elbows or ankles while retaining useful strength. Regular review can identify changes before they seriously limit movement.

Heart disease can progress separately from muscle weakness. A person who walks well and

has few physical limitations can still develop a significant rhythm problem. Lifelong cardiology follow-up remains important, including after a pacemaker or another device is fitted.

Relatives with the same genetic change may have different symptoms. Some female carriers of an EMD change have few muscle symptoms but remain at risk of heart disease.

Genetic conditions can also affect family planning, employment, independence, and worries about relatives. Muscular dystrophy organisations, genetic counsellors, rehabilitation teams, and mental health professionals can provide practical and emotional support.

When to get medical help

Call emergency services for:

These symptoms can have causes other than EDMD, but they need urgent assessment.

Seek prompt medical advice for new palpitations, near-fainting, worsening breathlessness, leg swelling, or a clear drop in exercise ability. Do not wait for a routine appointment if symptoms are worsening.

Routine follow-up should include planned heart monitoring even when you feel well. At regular appointments, mention increasing falls, pain, joint tightening, sleep-related breathing symptoms, or difficulty with daily activities so support can be adjusted.

- Fainting, especially during activity or with heart palpitations
- Severe or sudden shortness of breath
- Chest pain that is new, severe, or does not settle
- A sustained very fast or irregular heartbeat with dizziness
- Signs of stroke, such as facial drooping, arm weakness, or speech difficulty

Common questions

Is EDMD the same as Duchenne muscular dystrophy?

No. They usually involve different genes and patterns. Early contractures and heart rhythm problems are particularly characteristic of EDMD, and muscle weakness is often more slowly progressive than in Duchenne muscular dystrophy.

Can someone with EDMD still walk?

Many people walk for a long time. Contractures and weakness can eventually make this harder, and some people use braces, walking aids, or a wheelchair. Needs vary and may change over

time.

Can EDMD affect women?

Yes. Women can have EDMD caused by certain genetic changes. Females carrying an EMD change may have no obvious weakness but can still develop heart rhythm disease. Monitoring should be based on genetic and cardiac assessment.

Does everyone with EDMD need a pacemaker?

No. A pacemaker is used when testing shows a significant slow rhythm or electrical conduction problem. Some people need an ICD because it can also treat dangerous fast rhythms. Regular monitoring helps determine whether a device is needed.

Is exercise safe with EDMD?

Appropriate activity is often helpful, but it should match the person's strength, heart health, and joint movement. Moderate, low-impact activity is generally better tolerated than heavy lifting or repeated exercise to exhaustion. A physiotherapist familiar with muscle conditions can help plan safe exercise.

Can EDMD be passed to my children?

It can be, but the chance depends on the gene and inheritance pattern. Genetic testing and counselling can explain the result for your family and whether relatives may benefit from testing or heart screening.

Sources

- GeneReviews: Emery-Dreifuss Muscular Dystrophy , University of Washington, Seattle; hosted by the National Center for Biotechnology Information.
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