CONDITION

Childhood muscular dystrophy

Learn about childhood muscular dystrophy, including possible symptoms, genetic causes, testing, treatment, complications, and ongoing care.

WHAT IT IS
Group of genetic conditions causing progressive muscle weakness

BODY SYSTEM
Muscles and movement

MAIN SYMPTOMS
Delayed motor milestones, frequent falls, movement difficulties, progressive weakness

WHAT CAUSES IT
Genetic changes, sometimes inherited

HOW IT IS DIAGNOSED
Genetic testing, sometimes muscle biopsy

MAIN TREATMENT
Supportive multidisciplinary care

CAN IT BE CURED?
No, most forms have no cure

IS IT AN EMERGENCY?
Sometimes, with severe trouble breathing

LONG-TERM OUTLOOK
Progressive weakness with a widely varying course

Overview

Childhood muscular dystrophy is a group of genetic conditions that cause muscles to become weaker over time. The conditions affect proteins that muscles need to develop, function, or repair themselves.

There are many types of muscular dystrophy. Duchenne muscular dystrophy is one of the most common types beginning in childhood. Other forms include Becker, congenital, limb-girdle, Emery-Dreifuss, facioscapulohumeral, and myotonic muscular dystrophy. The age when symptoms begin, the muscles affected, and the rate of progression vary widely.

Some forms mainly affect movement. Others can also affect the heart, breathing muscles, joints, spine, learning, or behavior. A child may need care from several specialists even if they currently have only mild symptoms.

Muscular dystrophy is different from muscle weakness caused by an injury or lack of activity. It is also different from cerebral palsy, which results from changes or injury to the developing brain and usually does not cause progressive muscle damage.

There is not yet a cure for most muscular dystrophies. However, treatment can help maintain movement, protect the heart and lungs, manage symptoms, support learning, and improve quality of life. Some newer treatments target a specific genetic change, so an accurate diagnosis can affect treatment choices and eligibility for clinical trials.

Symptoms

Symptoms depend on the type of muscular dystrophy. They may appear during infancy, early childhood, adolescence, or later. Some children develop slowly at first, while others meet early milestones and then begin having difficulty with activities they previously managed.

Possible movement-related signs include:

A child with weakness around the hips may use their hands to "walk" up their thighs when standing from the floor. Clinicians call this Gowers sign. It can occur with muscular dystrophy, but it is not enough by itself to identify the cause.

Some forms can cause low muscle tone, known as hypotonia, during infancy. A baby may seem unusually floppy or have difficulty holding up their head. Feeding, swallowing, or breathing problems may also occur in certain congenital forms.

Symptoms outside the muscles may include an abnormal heart rhythm, reduced heart-pumping strength, breathing problems during sleep, or curvature of the spine. Some children with dystrophin-related muscular dystrophy have learning, attention, language, or behavioral differences. These concerns do not affect every child and are not a measure of intelligence.

- Delayed sitting, standing, walking, or other motor milestones
- Frequent falls or difficulty keeping up with other children
- Trouble running, jumping, climbing stairs, or rising from the floor
- Walking on the toes or developing an unusual walking pattern
- Weakness in the hips, thighs, shoulders, arms, face, hands, or lower legs
- Enlarged calf muscles caused by muscle tissue being replaced by fat and connective tissue
- Muscle stiffness, cramping, pain, or unusual tiredness

- Tight joints, also called contractures
- Loss of movement skills over time

Causes and risk factors

Muscular dystrophy is caused by a disease-related change, or variant, in a gene. Different genes provide instructions for different muscle proteins. When one of these proteins is missing or does not work correctly, muscle fibers are more easily damaged.

The inheritance pattern depends on the type. Duchenne and Becker muscular dystrophy are usually X-linked. They most often cause symptoms in boys, although girls and women who carry a dystrophin gene variant may develop muscle or heart symptoms.

Other muscular dystrophies may be inherited in an autosomal recessive pattern, meaning a child receives a changed copy of the gene from each parent. Autosomal dominant forms can result from one changed gene copy inherited from either parent.

A family history may increase the chance of an inherited muscular dystrophy. However, a child can have the condition without a known family history. A genetic change may be new in the child, or relatives with the change may have mild or unrecognized symptoms.

Muscular dystrophy is not caused by parenting, exercise, diet, infection, or something a child did. Routine physical activity does not cause the genetic condition, although exercise plans should be adapted to avoid excessive muscle strain.

Diagnosis

Evaluation usually begins with a medical and family history and a physical examination. The clinician may assess muscle strength, reflexes, walking, balance, joint movement, and how the child rises from the floor. Development, school performance, breathing, and heart symptoms may also be reviewed.

A blood test can measure creatine kinase, or CK. This enzyme leaks into the blood when muscle is damaged. A very high CK level can support concern for a muscle disorder, but it does not identify the exact type. CK can also rise after strenuous activity, injury, or other muscle problems.

Genetic testing is central to diagnosis. It may look first for changes in a specific gene, such as the dystrophin gene, or use a panel that examines many genes linked to muscle disease. Broader testing may be considered when earlier results are unclear. Results should be

interpreted with the child's symptoms and examination because some genetic findings have uncertain meaning.

Other testing may include:

A pediatric neurologist or neuromuscular specialist often coordinates the evaluation. Genetic counseling can help families understand test results, inheritance, testing options for relatives, and future pregnancy considerations.

- An electrocardiogram and echocardiogram to check heart rhythm and function
- Breathing tests, sleep studies, or blood oxygen measurements
- Imaging of muscles with MRI
- Electromyography, which evaluates electrical activity in muscles and nerves
- A muscle biopsy when genetic testing does not provide a clear answer

Treatment and management

Treatment is based on the specific type of muscular dystrophy, the child's age, symptoms, genetic result, and overall health. Care is often provided through a multidisciplinary neuromuscular clinic.

Physical and occupational therapy can help preserve comfortable movement and participation in daily life. Stretching, positioning, braces, and splints may help delay contractures. Occupational therapists can recommend ways to make dressing, writing, bathing, eating, and school activities easier.

Regular physical activity is generally encouraged when it is adapted to the child. Gentle, low-impact activities such as swimming or assisted cycling may support mobility and general health. Exhaustive exercise and heavy resistance work may damage already vulnerable muscles. A physical therapist familiar with neuromuscular conditions can help develop a suitable plan.

Mobility equipment is not a sign that treatment has failed. Scooters, wheelchairs, standing devices, and transfer equipment can conserve energy, reduce falls, protect independence, and help a child participate at school and in the community.

Corticosteroid medicines are commonly used in Duchenne muscular dystrophy because they can slow loss of strength and movement. They may also support breathing and heart function. These medicines require ongoing monitoring because they can affect growth, weight, mood, blood pressure, bones, blood sugar, and the body's response to physical stress. Families should not start, change, or suddenly stop a corticosteroid like prednisone or hydrocortisone without

guidance from the treating team.

Some gene-based medicines are available for selected children with Duchenne muscular dystrophy. Eligibility depends on the exact genetic variant, age, health status, and the medicine's authorization criteria. These treatments do not apply to every child, and their possible benefits, uncertainties, and monitoring requirements should be discussed with a neuromuscular specialist.

Heart care may begin before symptoms appear. A cardiologist may use medicines to protect heart function or treat reduced pumping strength and abnormal rhythms. Breathing care may include assisted coughing, airway-clearance techniques, treatment of infections, and noninvasive ventilation delivered through a mask, especially during sleep.

Nutrition support should balance growth, bone health, swallowing safety, bowel function, and the risk of unwanted weight gain or loss. A swallowing assessment may be needed if meals take a long time or cause coughing or choking.

School support can include physical access, extra time between classes, help carrying materials, rest periods, adaptive technology, and assessment for learning or attention needs. Emotional and social support are also important for the child, siblings, and caregivers.

Complications

Complications vary by diagnosis and may develop gradually. Weak breathing muscles can lead to shallow breathing during sleep, ineffective coughing, or repeated chest infections. Heart muscle disease can reduce pumping strength, while electrical changes may cause an abnormal rhythm.

Muscle imbalance can lead to contractures, hip problems, or scoliosis, which is a sideways curve of the spine. Reduced weight-bearing, limited mobility, delayed puberty, and corticosteroid treatment can contribute to weak bones and fractures.

Swallowing weakness may cause poor nutrition or allow food and liquid to enter the airway. Reduced mobility can also contribute to constipation, pain, pressure injuries, and loss of independence.

Regular screening matters because heart or breathing changes may be present before a child notices symptoms. Early support can reduce the effect of some complications.

Follow up

Routine follow-up usually includes assessments of strength, movement, joint flexibility, growth, nutrition, bone health, heart function, breathing, sleep, and emotional well-being. The timing of visits and tests depends on the type of muscular dystrophy and the child's current needs.

Contact the care team promptly for a clear decline in walking or daily function, new swallowing difficulty, increased falls, persistent pain, morning headaches, unusual daytime sleepiness, or signs of a chest infection. These concerns may require an earlier assessment but are not always emergencies.

Seek urgent or emergency care for:

Families should keep an updated list of the diagnosis, medicines, allergies, specialists, breathing equipment, and emergency instructions. Before surgery, anesthesia, or sedation, tell the medical team about the muscular dystrophy. Planning may require input from neuromuscular, heart, breathing, and anesthesia specialists.

- Severe trouble breathing, blue or gray lips, or inability to speak because of breathlessness
- Choking with ongoing breathing difficulty
- Fainting, severe chest pain, or a sustained rapid or irregular heartbeat
- Sudden major weakness, especially with fever, dehydration, or dark-colored urine
- A serious fall, suspected fracture, or new inability to move a limb

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