Duchenne muscular dystrophy (DMD)

What is Duchenne muscular dystrophy or DMD?

Muscular Dystrophy is a group of inherited muscle disorders, in which muscles weaken over time. Duchenne muscular dystrophy is the most common form of muscular dystrophy. It occurs in about 1 out of 3,500 live male births.

Duchenne Muscular Dystrophy is caused by a change (mutation) of the gene that makes a protein called dystrophin. This protein is needed for muscle cells to keep their shape and work properly. An abnormal gene cannot make this protein and without it, muscle cells collapse and die. This causes the symptoms of DMD to develop.

What are the signs and symptoms of DMD?

The first symptoms of DMD usually appear between 2 to 5 years of age.

Symptoms of DMD may include:

- Muscle weakness – The first muscles that are affected are those around the hips and upper thighs. Children may have difficulty walking, running, jumping, climbing stairs or standing up from the floor.
- Loss of muscle coordination – Children may seem unsteady, clumsy, fall often or “waddle” when they walk.
- Muscle size – As the muscles are damaged, the muscle is replaced by fat and connective tissue and can shrink (atrophy). The calf muscles often become larger (hypertrophy) and other muscles may be enlarged early on.
- Toe walking – The muscles become tight around joints, limiting movement. When walking becomes difficult, children may walk on their “tippy-toes” or balls of their feet.
- Learning disabilities – Some children have learning difficulties, which are usually not severe and usually do not get worse.

DMD is a progressive disorder, meaning the muscles get weaker over time. The muscle weakness spreads and begins to affect the ability to walk. Later on, muscles of the heart and those around the lungs become affected.

How do you know my child has DMD?

Your doctor or specialist can tell if your child has DMD from:

- talking with you about your family history
- examining your child for signs and symptoms
- reviewing the results of the following tests

Blood tests

- CK level – Muscles that are weak or damaged leak an enzyme called CK (creatinine phosphokinase) into the blood. High levels of CK in children can be a sign of DMD or another muscle disease.
- Genetic tests – Checking a sample of blood for the abnormal gene for DMD.

Muscle biopsy

- A biopsy is a procedure to take a tiny sample of muscle to be examined under a microscope.
- A biopsy is only needed if the first genetic test returns with a negative result. This occurs in about one-third (30%) of cases.
- The procedure is done in the clinic and takes about 20 minutes. The sample is usually taken from the thigh muscle (quadriceps). First the area is ‘frozen’ or numbed with a local anesthetic. Then the doctor makes a tiny cut (incision) and removes a sample of muscle with a needle. The incision is closed with a stitch that will need to be removed in a few days.

How did my child get DMD?

DMD is inherited. The abnormal gene for DMD is found on the X chromosome, passed down from the mother.

- In about 66% of the cases, the mother carries the disease-causing gene in all the cells of the body (see below) and will need to be tested to see if she does carry the gene mutation.
- In about 33% of the cases, the mutation is only in the ovary (germline mosacism) or randomly occurs during the development of the embryo (spontaneous mutation).

How boys are affected

- Boys have one X chromosome inherited from their mother and one Y chromosome inherited from their father.
- If a boy inherits an X chromosome with the abnormal gene for DMD from his mother, he will develop the disorder.

How girls are affected

- Girls have two X chromosomes.
- If a girl inherits an X chromosome with the abnormal gene for DMD, the other X chromosome has a normal copy of the gene. The normal gene can usually make enough protein to prevent symptoms.
- Girls are not usually affected by DMD. They can be carriers, meaning they have one abnormal gene, but not the disorder. A woman carrying the DMD mutation can develop mild muscle weakness and an enlarged heart later in life. This happens when the normal copy of the X chromosome does not fully protect her from the abnormal gene.
- If a woman carries the abnormal gene for DMD, each of her children will have a 50% chance of inheriting the X chromosome with the abnormal gene.
  - If a son inherits the abnormal gene, he will develop DMD.
  - A daughter inheriting the abnormal gene will become a carrier.

How is DMD treated?

The health care team at the Neuromuscular and Neurometabolic Centre will recommend a treatment plan to meet your child's needs. The goals of treatment are to control symptoms, keep muscles working as long as possible and give your child the best possible quality of life. There is no cure for DMD.

Physiotherapy

- It is important for your child to be active. Using muscles helps them stay as strong as possible. Inactivity, such as bedrest, can cause the muscles to break down faster.
- A physiotherapist can design a daily program of stretches, guided exercise and activity to suit your child's age and ability. This will help keep muscles and joints moving well for as long as possible. Swimming and exercising in water can be helpful as these activities do not put a lot of stress on joints.
- Your child should not "over do it". Exercising strenuously or for a long time can hurt your child's muscles. It is important for parents not to push exercises such as heavy weight lifting. Help your child "listen" to his or her own body and never push to the level of damaging the muscles.

Corticosteroids

- Steroid medications can slow the progress of the disorder. It is important to remember that side effects are possible with all medications.
- Prednisone can help keep muscles strong and working well. Deflazocort works in the same way as prednisone, but may have fewer side effects. Deflazocort may not be covered by your drug benefit plan.

Assistive devices

- Orthotics, braces, canes and walkers can help your child walk and do things for him/herself.
- Later on, a wheelchair can help your child get around.

Orthopedic treatment

- As the disorder progresses, tendons and joints can become tight and the bones in the back and chest can become deformed. Your child may visit an orthopedic specialist for treatment, including surgery.

Treatments to help with breathing

- Muscle weakness and changes in bones contribute to problems with breathing. BiPAP and other machines can help make breathing easier in the later stages of the disorder.

Treatments to help the heart

- DMD can lead to high blood pressure and heart problems such as a thickening of the heart wall (cardiomyopathy).
- The doctor may prescribe medications to lower blood pressure and reduce the work of the heart.

When should my child see the doctor?

Follow-up visits

- The health care team will regularly assess your child and adjust the treatment plan. This means your child will have regular follow-up visits at the Neuromuscular and Neurometabolic Centre as well as with other specialists.
- Your child will also need regular tests to check his or her heart, lungs and muscle strength.

When to get medical help

- **Call the specialist if your child's symptoms get worse or new symptoms develop.**
- **If your child has difficulty breathing, call 911 or go to the nearest hospital emergency room.**

What is the prognosis with DMD?

By late childhood or early teen years, some children are unable to walk. The average age when a child loses the ability to walk independently is 12 years.

Sadly, nothing can stop the progression of DMD. Symptoms become worse over time, leading to increasing disability. The best possible treatment may delay the average time until wheelchair use by 2 years. By the mid 20's, the disorder is severe enough to cause death, usually as a result of complications with breathing.

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What help is available for my family?

Support for your family

- You, your child and your family are facing many challenges - physical, emotional and financial. It can help to have a support system (for example, family, friends, teachers, religious or spiritual leaders) around you. You may also find it helpful to talk with other families who are coping with a similar situation.
- The hospital social worker can also help you and your family find ways to cope.
- Another source of support is the Muscular Dystrophy Association.

Genetic counselling

- If you have a family member with DMD, speaking with a genetic counselor may be helpful.
- During an appointment with your specialist, you can talk about carrier status, monitoring and testing of family members.
- If the gene for DMD is identified, prenatal testing early in pregnancy can detect if an unborn baby has the disorder.

If you have any questions about Duchenne muscular dystrophy or your child's care, please speak with your doctor or nurse at the Neuromuscular and Neurometabolic Centre.

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