

DUCHENNE MUSCULAR DYSTROPHY: A GUIDE FOR PARENTS



Duchenne muscular dystrophy: A Guide for Parents

This information was originally developed for families of children recently diagnosed with Duchenne Muscular Dystrophy (DMD) by Bloorview Kids Rehab - Neuromuscular Team. It outlines the cause of the disorder, treatments, current research, and other issues important for families. Further information is available from Bloorview Kids Rehab - Neuromuscular Team, The Hospital for Sick Children - Division of Neurology, and Muscular Dystrophy Canada.

What is Duchenne muscular dystrophy?

DMD is an inherited disorder. The muscles become weaker as the boys get older. This is because the body can not make the muscle protein called dystrophin. This makes the muscle cells weak and they gradually break down. DMD usually affects boys (it is very rare in girls). Signs of weakness start when the boys are between 3 and 5 years of age (sometimes earlier). At first, the weakness is seen mostly in the legs and hips. The children may:

- Fall frequently
- Have trouble running as fast as their friends
- Have trouble climbing stairs
- Have trouble getting up from a chair
- Develop big calves
- Frequently walk on their toes and lean backwards to keep their balance
- Eventually this weakness also makes walking more difficult and a wheelchair is needed. Gradually all the muscles become very weak including the muscles used for breathing and the heart.

Can We Be Sure There is No Mistake in the Diagnosis?

The doctor was able to confirm that your son has DMD because:

- He has muscle weakness and large calves usually seen in muscular dystrophy.
- The muscle enzyme in his blood called CK is very high. This means the muscle cells are breaking down.
- His DNA blood test (genetic test) showed a change in his dystrophin gene, if the genetic test was normal, his muscle biopsy showed a lack of dystrophin in the muscle cells.

What causes Duchenne muscular dystrophy?

The cells in our body contain a list of instructions stored as units of information called genes. Within each cell there are thousands of different genes. These genes are responsible for building and maintaining our body. One of these genes is the dystrophin gene. It is responsible for making the protein dystrophin which keeps muscles strong and

healthy. DMD is caused by a change in the dystrophin gene. This change is referred to as a mutation. When there is a mutation in the dystrophin gene, the protein dystrophin is not made properly. The muscle cells become weak and they gradually break down.

How Do Mutations Happen?

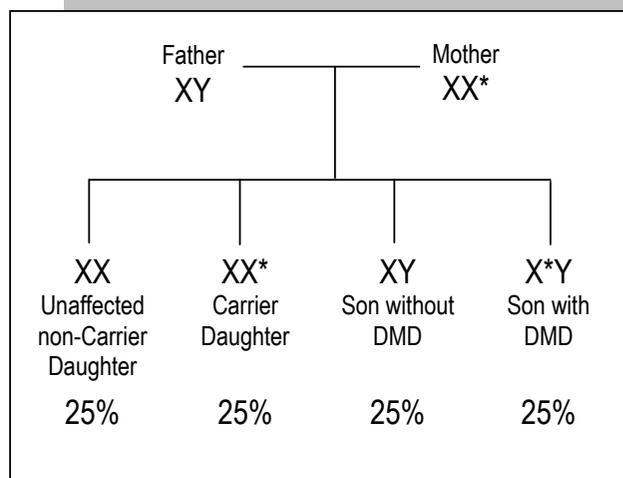
We do not know why mutations happen. They just happen sometimes in all living cells. Everyone has a few mutations in their genes which do not cause problems. However, in some cases a mutation can lead to a disease or chronic condition like DMD. No one is at fault when a mutation happens; it is not the result of anything anyone has done.

How is DMD Inherited?

Approximately 1/3 of the time, the mutation in the dystrophin gene comes as a "new change" for the gene. The rest of the time, the DMD mutation can be passed down from mother to son. This is natural, and it is not anyone's fault.

Chromosomes are the structures that contain our genes. Girls have two X chromosomes (one from her mother and one from her father). Boys have one X chromosome (from his mother) and one Y chromosome (from his father). The dystrophin gene is located on the X chromosome. If the mother passes on the X chromosome with an altered dystrophin gene to her son, he will develop DMD. Girls who inherit the DMD mutation from their mothers also inherit a normal copy of the DMD gene on the X chromosome from their fathers. These girls will be carriers. They don't have DMD but like their mothers, they can pass DMD on to their sons.

Every time a mother who is a carrier of DMD becomes pregnant there are four possible outcomes. These are illustrated below:



X* = X chromosome with mutation in the dystrophin gene.

What Does It Mean to be a 'Carrier' of DMD?

A woman who is a carrier of DMD has one X chromosome with the altered dystrophin gene, and one normal X chromosome. Some women who have carrier status experience increasing muscle weakness and fatigue. These are known as carrier symptoms. All women who are carriers should have their heart checked regularly as the heart muscle can be affected.

What if We Want to Have More Children?

You are encouraged to seek genetic counseling to talk about risks and options before you get pregnant. If you are pregnant, testing may be available to tell if your unborn son has DMD.

How Can a Genetic Counsellor Help?

Genetic counselors are available to review and assess your family history. This helps you understand the genetics and inheritance of DMD. They can discuss genetic testing and review your choices for prenatal testing.

What Can Be Done to Help?

There are several different ways you can help maintain your son's muscle strength:

1. Physiotherapy can help by:

- Giving you information about avoiding over exercising
- Maintaining correct foot position by means of:
 - a) daily ankle/foot stretches which you can be taught to do at home
 - b) use of below knee braces (if these are appropriate)
- Counselling about recreation and leisure activities which will help maintain your child's abilities.

2. Medication

Deflazacort

A steroid medication, Deflazacort, can help preserve muscle strength for some time. It is now used by many boys with DMD. However this medication has significant side effects. Some families choose not to use it.

Boys who take Deflazacort are usually able to continue walking for a much longer period, some until they are 12 years old or more.

Boys generally start taking steroid medication when they show some signs of muscle weakness. This could include: walking on their toes; increased difficult climbing stairs; and falling more often. This often occurs when the boys are 6 - 8 years old.

Steroid use and its effects will be fully explained to you. Your child will be closely monitored for possible side effects.

Alendronate Medication

One of the side effects of Deflazacort use and lack of physical activity – such as sitting in a wheelchair - is loss of calcium in the bones (osteoporosis). This may increase the risk of fractures. Alendronate has successfully helped osteoporosis in adult women.

Are There Any Medications to Avoid?

Boys who have DMD may not react well to the anesthetic agent succinylcholine. It is recommended that boys wear a "Medic Alert" bracelet, which states their diagnosis and whether they use steroids (Deflazacort). This will alert any health care professional to avoid the use of succinylcholine during surgery.

3. Surgery

Boys with muscle weakness develop stiffness in some joints - such as the ankle - and curvature of the spine (scoliosis), usually in early adolescence. Surgery can correct these conditions.

Should Other Members of My Family Be Tested For DMD?

Sons

If you have another son who has signs of muscle weakness, a doctor who is a neurologist should check him.

Daughters

If you have a daughter, she should be told about the possibility of being a carrier. You can do this when you think she is old enough to understand. She should get genetic counseling and carrier testing before planning a pregnancy.

Sisters of Carrier Mothers

If genetic testing shows that the mother of a boy with DMD is a DMD carrier, then her sisters may also be DMD carriers. They should be informed of this risk and encouraged to seek genetic counseling, particularly if they are planning to have children. A neurologist should assess any of their sons who show signs of muscle weakness.

4. Maintenance Of a Healthy Lifestyle

This will help your son to stay as healthy as possible. Things that may help include:

- a) Involving your son in recreational, leisure activities. These will vary depending on your child's interests and abilities but may include swimming, cycling (with or without adaptations), specialty skiing, Tae Kwon Do, wheelchair hockey, etc.
- b) Provision of an accessible environment at home and at school. Stairs are difficult for boys with muscle weakness. One storey houses and schools, or schools with elevators are recommended.
- c) Maintaining healthy lungs. This is important as muscle weakness affects the muscles used for breathing. A smoke free environment, use of flu vaccines, regular assessment of lung function at clinic visits, - are all important.

5. Ventilation

This is a choice of treatment which can help or replace the ability to breathe. It can be used later in life when the muscles used for breathing become weak.

Are There Any Other Things Which Can Help?

You may hear about several other 'treatments' for DMD. Some of these are controversial and their effectiveness has not yet been proven. We will discuss any 'treatment' with you. These alternative treatments include the following:

Myoblast Transplants

In this procedure donor cells are injected into certain muscles in the hope that they will fuse with diseased muscle so that the muscle fibres will contain some normal muscle which will produce the dystrophin gene. So far this treatment has not been effective.

Other Alternative Therapies

There are many alternative therapies such as chiropractic treatments, nutritional supplements, and naturopathic preparations which some people use. None of these provide a cure but many families believe they help with some aspects of the disorder.

How Will DMD Affect My Child at School?

Some boys who have DMD have learning difficulties. This may not be noticed until they start school. Special testing to determine specific learning needs is available. These learning difficulties do not become worse over time. Stairs in the school may also be a problem. Your child may need to change to a more accessible school. The neuromuscular Team recommends the development of close liaisons between you, the Neuromuscular Clinic and your child's school in order to identify and address any areas of concern.

Is There a Cure for DMD?

We do not have a cure for DMD at this time, but there is a tremendous amount of research currently taking place in many countries. Researchers have made great advances in their knowledge of DMD. They are currently working in several areas which they believe will eventually produce a cure. Some of these areas are:

Gene Therapy

Or, more accurately, gene replacement is one of the most promising areas. Scientists have developed a synthetic gene which they hope to introduce into the body to 'take over' and produce the missing protein dystrophin. At present there are several difficulties to be overcome before this can be tried as a treatment for DMD.

What Should I Tell My Child?

Talking to children about DMD may be difficult, but it is important for them to have some information about their (or their brother's) condition. What you tell children will depend on their age and level of understanding.

For instance, for young children, an explanation about being "born with weak muscles" may be enough. Children need some information or they may become confused and frightened, even blaming themselves for the problems. Talking with your children about DMD will help them accept themselves and the physical limitations association with DMD.

***Produced by Bloorview Kids Rehab,
Neuromuscular Team
Adapted with permission by Muscular
Dystrophy Canada
Updated: September 2007***

How Can We Cope With This Diagnosis?

A diagnosis of DMD comes as a shock, and brings challenges and difficult adjustments for a family. At first you may experience many mixed emotions, especially grief, anger and fear. Giving yourself time to adjust, talking to family, friends and professionals, and reading about DMD, may help as you deal with this diagnosis. Talking to other parents of children with DMD may also be helpful. Families who have older children with DMD may also be helpful. Families who have older children with DMD can tell you that in time you can adjust and continue to lead a good life again.

Children with DMD will experience physical limitations as they grow and their condition changes. Even so, they are children and like other children, need to have as normal a life as possible. Developing self-esteem, having fun, participating in recreational activities, and living a normal life with family and friends at home, at school and in the community.

How can I help?

Muscular Dystrophy Canada conducts year-round fund raising campaigns to support our diverse programs. Your gift will help the Association provide the dollars necessary to assist individuals living with neuromuscular disorders, and fund much needed medical research and educational information. Please make a gift through our National office or any Regional or Community Muscular Dystrophy Canada office.

**All Muscular Dystrophy Canada Information
Sheets are available on our website:
www.muscle.ca**

**Ce feuillet d'information est aussi disponible en
français.**

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