Frequently Asked Questions

1. **If no members of my family are affected by DMD, can I still have a child with the condition?**
   Yes. Due to the way the disease is inherited, it is possible for the defective gene to be present in the family without anyone showing symptoms. Hence, you may not be aware of a family history. In addition to that, there is also a 1/3 risk of spontaneous mutations occurring in families with no history of the condition.

2. **If I am a carrier, can I develop DMD?**
   A minority of carriers (females) have a mild form of the disorder. It is wise to have regular strength evaluations and cardiac monitoring to manage any symptoms that may arise.

3. **Why don't girls usually have DMD?**
   A girl has two copies of the X chromosome. Hence, when she inherits a flawed dystrophin gene from one parent, she usually also gets a healthy dystrophin gene from her other parent, giving her enough of the protein to protect her from the disease.

4. **If I already have one affected son, does that mean that I will not have another?**
   No. There is a 50% chance of having an affected son with each pregnancy.

5. **Is the muscle deterioration painful?**
   No. It is not painful in itself. Some people report muscle cramps at times, which can be treated with over-the-counter pain relievers.

6. **What other investigations/tests need to be performed on a DMD patient?**
   Complete and regular cardiac and respiratory evaluations.

7. **Since DMD is due to a protein deficiency, will eating more food with protein help?**
   No. Protein from food cannot replace the lost dystrophin.

References

Fact Sheet 41: Duchenne and Becker Types of Muscular Dystrophy [online]
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Facts About Duchenne & Becker Muscular Dystrophies. Muscular Dystrophy Association Inc. 2009

NORD Guide to Rare Disorders. Lippincott Williams & Wilkins. 2003

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MALAYSIAN RARE DISORDERS SOCIETY

Rare Disorders Series:

Duchenne Muscular Dystrophy

What is Duchenne Muscular Dystrophy (DMD)?

DMD is an inherited muscle disorder characterised by a rapidly worsening muscle wasting and weakness.

It is caused by a change (mutation) in the *DMD* gene, resulting in the failure in producing the Dystrophin protein.

Genes are located on chromosomes and contain 'recipes' to make proteins.

The *DMD* gene, which produces the Dystrophin protein, is located on the X chromosome.

Dystrophin is one of several proteins located along the membrane of muscle fibres. It functions to keep muscle cells working properly.

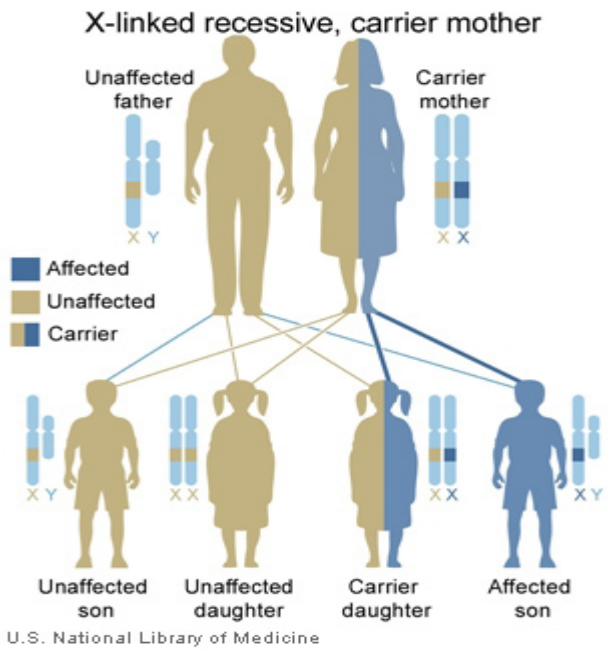
Because of the way the disease is inherited, males are more likely to develop symptoms than are females.

The sons of females who are carriers (women with a gene change, but no symptoms themselves) each have a 50% chance of having the condition.
The daughters each have a 50% chance of being carriers.

The rate of disease progression is variable. Symptoms usually appear before age 6.

Many children with DMD lose the ability to walk sometime between ages 9 and 13.

Patients are advised to consult a geneticist to obtain further information on disease progression.



Inheritance pattern of DMD

Signs and symptoms

- Late in learning to walk
- Enlarged calf muscles (pseudohypertrophy)
- Clumsy and falling frequently
- Trouble climbing stairs, getting up from floor and running
- Walking on toes or balls of feet
- Waddling and unsteady gait
- Difficulty raising arms

Other ways DMD may affect the body

- Abnormalities in heart muscles
- Chest and back deformities (scoliosis)
- Loss of muscle mass (wasting)
- Muscle contractures in knees and legs
- Muscle deformities
- Learning difficulties
- Respiratory disorders i.e. pneumonia and aspiration of food or fluid into the lungs

Testing

Genetic counselling by qualified personnel is advised if there is a family history of the condition and before any genetic testing is done.

- CK level
  To check the level of an enzyme called Creatine Kinase that leaks out of damaged muscles.

- Genetic
  To diagnose the type of muscular dystrophy affecting or carried by an individual by looking at the dystrophin gene mutation.

- Muscle biopsy
  May be performed when no mutations are found in the gene.

Treatment

There is currently no known cure for DMD.

Treatment is given to slow the progression of muscle weakness and to prolong mobility to some extent.

Steroid is the drug administered to help increase strength, muscle function and pulmonary function under certain circumstances with the guidance of the doctor.

Exercise and physiotherapy may be helpful to alleviate or mitigate contractures (fixations of the joints) which usually affect the knees, hips, feet, elbows, wrists and fingers.

Orthopaedic appliances such as braces, standing frames and wheelchairs may improve mobility and the ability to care for oneself.