

WHAT IS MUSCULAR DYSTROPHY?

Muscular Dystrophy is a genetic disease characterized by the progressive weakness and degeneration of the voluntary muscles of the body. The muscles of the heart and lungs are also affected during the later stages of the disease.

There are many forms of Muscular Dystrophy that affect children and adults.

- **Duchenne Muscular Dystrophy** is the most common childhood form and the one most frequently encountered in the school setting. This is the one we will be discussing in this booklet.
- **Becker Muscular Dystrophy** is the second most common form. It has signs and symptoms similar to Duchenne but generally appear later, progress more slowly and is generally less severe.

WHAT IS THE CAUSE?

Muscular Dystrophy is caused by a defect in the genes for muscle proteins. **Muscle proteins** act like glue supporting the structure of muscle fibers. **A gene acts like a recipe for the protein.** If the recipe is wrong, the protein is either created defectively, in the wrong amounts or sometimes not at all.

- In Duchenne or Becker Muscular Dystrophy, a muscle protein called **dystrophin** is either missing, deficient or abnormally formed.
- In the absence of these healthy muscle proteins, muscle tissue begins to degenerate.
- Degeneration may progress at different rates in different children depending on the genetic code that is passed down.

MUSCULAR DYSTROPHY IS AN INHERITED DISEASE

Inheritance is sex linked and passed down through the mother to her son. The defect lies with the X chromosome. Males have an XY chromosomal combination and females have an XX combination. **Girls do not inherit the disease** because only one of the X chromosomes would be affected, leaving one healthy to override the unhealthy one. On the other hand, **males inherit the disease** because they carry only one X chromosome. If it is defective then, they inherit the disease.

- Male children from a woman who is a carrier have a 50% chance of having the disease.
- Female children from a woman who is a carrier have a 50% chance of becoming carriers.

Muscle strengthening exercises are not recommended as they can actually speed up the degeneration process. However it is important to maintain a normal level of activity as long as possible.

What are the signs of MD?

Early signs of Duchenne Muscular Dystrophy occur between the ages of 2 and 6 years old. Prior to this parents are generally unaware of the disease process. Symptoms are as follows:

- Weakness first appears in the pelvis, upper arms and upper legs.
- Difficulty rising to a standing position from the floor.
- He may fall frequently.
- He may have a waddling type of walk.
- There is an apparent enlargement of the calf muscle caused by the accumulation of fat and connective tissue in the muscle.
- The child has difficulty climbing stairs.
- Scoliosis is a common result of spinal weakness later in the disease and may require surgery.
- Breathing is affected in the later stages of the disease.
- Boys with MD usually live into their late teens or early twenties.

Progression varies slightly from child to child. Frequently a wheelchair will be needed between the ages of 10 and 12 years of age.

Diagnosis is made through examination of a muscle biopsy and by checking the level of creatine kinase (CK) in the blood. CK is an enzyme that leaks out of damaged muscle, accumulating in the bloodstream.

What are the implications for Educators?

- **Mild** cognitive delay has been noted in approximately 10% of boys with MD.
- Aside from the obvious difficulties in mobility, these children learn much the same as normal children.
- As the disease affects their arms more, assistive technology or adaptation is useful to decrease fatigue and improve efficiency in assignment completion. (contact OT)
- Reducing the quantity of written work to create a more successful environment.
- Implementation of oral presentation, dictation or computer use to **maintain student participation**.

The Role of OT and PT in the Educational Setting

Physical Therapy:

- Act as a consultant to insure accessibility and independent mobility.
- Instruct on safe transfers in and out of a wheelchair.
- Maintain good positioning in wheelchair.
- Training for staff and student in power mobility when it becomes necessary.
- In-service training for educators and students on Muscular Dystrophy.

Occupational Therapy:

- Consultation on writing and fine motor performance as needed including accommodations or modifications for functional performance in these areas.
- Assistance with self-care tasks at school.
- Consultation on implementation of appropriate assistive technology that may be needed as the disease progresses.

Reference: *Facts about Muscular Dystrophy*: Muscular Dystrophy Assoc.

Further Resources:

1. <http://www.kidshealth.org>
2. <http://www.ldinfo.org>