Duchenne and Becker Muscular Dystrophy

➢Duchenne and Becker muscular dystrophy (BMD) are genetic disorders affecting a protein important to the structural integrity of the muscles, causing muscle weakness and deterioration, leading to varying degrees of disability and in many cases, to premature death.

➢Duchenne muscular dystrophy (DMD) is the most common type of muscular dystrophy (MD), affecting between 1 in 3,000 and 3,500 male births with an estimated overall prevalence of 63 per million people in the population. Persons from any racial or ethnic group may be affected.

➢Duchenne symptoms usually appear anywhere between 18 months and 4 years of age.

➢DMD Symptoms: Duchenne muscular dystrophy is marked by a childhood onset with progressive muscular weakness, starting in the lower legs and moving up, eventually involving the neck, shoulders, and arms. Often the first symptom seen in male children aged 3-6 years is an unusual "waddling gait," caused by pelvic girdle muscle weakness. Due to lower back and leg weakness, parents often see a boy pushing down on his knees in order to stand up (known as the Gower sign). Characteristic enlargements of the calf muscles are caused by degenerative changes in dead muscle fibres. Eventually the heart and muscles of respiration are involved, leading to various complications and eventually, death, usually in the early twenties.

➢Duchenne follows a typical course. The mean age of walking is about 18 months but an unusual gait is often obvious. Progressive weakness continues and by around the age of 8, most patients have difficulty ascending stairs and respiratory muscle strength begins a slow but steady decline. Obesity is a common feature. A wheelchair is usually required by about age 10 or 12 in most cases. Common complications can include: skeletal deformities, chest and back abnormalities (scoliosis), muscle deformities and severe progressive contractures, especially of the heels and leg. Death from heart and or respiratory complications usually occurs between 18 to 25 years of age; even with assisted ventilation, survival beyond the late twenties is rare.

➢Some 30% of Duchenne patients display mild but non-progressive mental retardation.

➢A continuum of severity of symptoms is observed; early onset and more severe cases are classified as DMD. Cases that typically present with a later onset and with milder symptoms are classified as Becker muscular dystrophy (BMD).

➢Becker muscular dystrophy tends to be more variable and typically shows a later age of onset (although it may range from childhood to mid-adulthood). Disability tends to be milder, for example, a wheelchair may not be needed until later in life, if at all. Lifespan may be reduced (average age of death is in the forties) but some patients live a normal lifespan. Becker is less common than Duchenne, affecting about 1 in 5,000 male births with an estimated overall prevalence of 24 per million people.

➢Cause: Both disorders involve mutations of the dystrophin gene found on the X chromosome, however, the type, location and severity of the mutation vary. Different mutations have different

impacts on dystrophin protein production and function and thus, the subsequent impact on muscle function is variable. In many Duchenne cases, severe mutations significantly disrupt large or critical parts of the dystrophin gene, preventing the production of any measureable dystrophin protein. Becker pateints have some functional dystrophin, creating milder symptoms.

➢DMD and BMD are inherited in an X-linked recessive pattern; therefore, DMD and BMD almost exclusively afflict males. Females are typically asymptomatic carriers.

➢Afflicted males cannot pass the mutation to sons, but all of their daughters receive the mutation and become carriers. Carrier mothers pass it onto sons 50% of the time (who become afflicted) and pass it onto daughters 50% of the time (who become carriers). About 30% of cases are not inherited; they involve a new spontaneous mutation (that subsequently may be passed on).

➢In some rare cases involving skewed X chromosome inactivation, females can manifest DMD and BMD symptoms, in some instances, as severe as males.

➢Diagnosis: Symptoms of DMD and BMD might be confused with other muscular dystrophies, for example, some forms of Limb Girdle muscular dystrophy. Ideally, patients will be diagnosed based on fully characterized biochemical (protein) and molecular (genetic) analyses.

➢Genetic Testing: DNA tests can show the presence and size of the more serious forms of mutation (usually deletions and duplications) but are not as accurate in the case of the point mutations that affect some 30% of DMD and BMD cases. Prenatal testing is generally available.

➢There are currently no treatments for either DMD or BMD that can halt or reverse the symptoms and muscle weakness. Each symptom displayed reflects an underlying issue and each needs to have a treatment plan developed to best address the concern on an individual basis.

➢ANESTHESIA WARNING: A higher rate of complications is associated with general anesthesia in patients with neuromuscular disorders. Even mildly affected patients may have complications. Patients with DMD or BMD or a family history of DMD or BMD need to discuss this with their surgeons.

Neuromuscular disorders and what people commonly call muscular dystrophies are very complex and often devastating conditions. Some strike children, others do not display symptoms until well into adult life. Most are progressively disabling, getting worse over time and some are fatal, often in childhood. Some affect primarily muscles, others can affect a wide spectrum of health, including; personality and intellectual functioning, the eyes, the bones, digestion and diabetes to list just a few. There are over 100 different types of disorders and variations listed under this umbrella. Many disorders have genetic causes but in many cases, just how these genetic mutations cause the symptoms we see is not yet understood. Every day, medicine is making progress in understanding these disorders, but at the present time, there are more theories than facts. Sadly there are no direct treatments yet.

Please feel free to contact Muscular Dystrophy Canada if you have suggestions or questions about this page. Our website is: http://www.muscle.ca/ or e-mail us at info@muscle.ca

Thank you.