

Living With Duchenne Muscular Dystrophy

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Introduction

Duchenne Muscular Dystrophy, (DMD), is an inherited sex-linked recessive disease, in which females carry and transfer an abnormal gene onto their male offspring. According to Terri Metules DMD affects exclusively males, one in every 3500 births. Metules, T. (2002).

The purpose of this paper is to offer a better understanding of DMD, both from the technical aspect and from the most important human aspect, that is in the day to day struggles of a live human being suffering with the disease. I have interviewed a Survivor of DMD, Bill Watts. We will look at Bill's daily routine, his early stages of this disease and how it has progressed into his present life. I have also included supplemental research to explain how DMD effects the human body. To start off, we will review some information from Bill's interviews, starting off with his DMD diagnosis.

Presentation & Discussion

Bill's DMD Diagnosis

Bill Watts describes his disease, DMD, or Duchenne Muscular Dystrophy, as a genetic spontaneous mutation, which is progressive and gradually weakens the leg muscles first and then gradually attacks other areas such as the heart and lungs until failure occurs. Usually those diagnosed with DMD do not live past their 21st birthday. Bill is 40 years old and despite his condition, he has a positive outlook about life and is very active with his social activities and religious activities at Good Shepherd Raker Center. To get a better understanding of Bill's life and disease condition, we will look at his DMD progression.

Bills DMD Progression:

The Symptoms of DMD for Bill occurred at 4 years of age, in which he was becoming fatigued when running. Upon turning five he started having difficulty standing up from a sitting position. At ages 13 – 16 years, Bill was able to take care of basic needs, such as dressing, toileting, showering and needed a walker for walking assistance. He progressed into a manual wheelchair at age 13 and was able to propel himself. When he was in 10th Grade and now 15 – 16 years of age, he used a motor chair and it was at 21 yrs. old when he started needing assistance, which progressed and he was fully dependent on others between 24- 25 yrs. To get an idea of what Bill does to keep himself busy, we will next discuss his daily routine.

Bills Daily Routine:eat

I asked Bill, “What is your day to day routine like?” Bill communicated that he gets up at 8am or 9am. At 10am, he has respiratory treatments, takes his medications and throughout the day has suction and insuflation treatment, which is like coughing for us, but cleans out extra mucus that has accumulated in his respiratory tract. This is done about four times a day and or as an as need basis by the Nursing Staff. He then proceeds with his daily plans, in which he pursues other interests, participates in activities and Prayer time in the chapel. He eats two meals a day and snacks when he gets hungry. He prefers not to eat breakfast, since it takes time away from his activities. Next, I will explore DMD and what causes these drastic changes that occur in Bill's muscle cells, hence the restriction of his activities of daily living.

Etiology

The cause of DMD is due to changes that occur in Bill's muscle cells, which lack dystrophin. Dystrophin is a protein that links cytoskeleton to the extracellular matrix, which acts as girders to help stabilize the sarcolemma. Tremblay, J. (2006). Without this protein Bill's sarcolemma is fragile and tears during muscle contraction and allows excess Calcium (Ca²) to build up. This influx of calcium creates a homeostatic imbalance and damages the contractile fibers which then breakdown, and macrophages and lymphocyte accumulate in the surrounding

connective tissue. This reduces the regenerative capacity of the muscle and the damaged cells undergo apoptosis, (programmed cell death), resulting in decreasing muscle mass.

MarieB & Hoehn (2007).

How do you know if you have DMD? Below is a list of some symptoms of DMD. Of course, further tests are needed if these symptoms persist.

Symptoms of DMD

1. Becoming clumsy and falling frequently, due to leg muscles weakening.
2. Those with this disorder are late in learning how to walk. Standing up from a sitting position is difficult, as well.
3. Toddlers may experience pseudohypertrophy, which is enlarged calf muscles.
4. Children may walk with a rolling gait, using the balls of their feet. In order to keep balance, the child will stick out their belly and put their shoulders back.
5. Difficulty raising arms. MDA (2007)

Gender Relationship

Duchenne muscular dystrophy, DMD, is the most common and serious form of muscular dystrophy. This disorder affects predominately boys between the age of two and seven years old. Bill was diagnosed between the age of four and five. Bill had symptoms of DMD. To determine an official diagnosis, the doctors had to run certain diagnostic tests. MarieB & Hoehn (2007).

Diagnostic Tests

In order to determine which kind of muscular dystrophy Bill had, the doctor ordered a muscle biopsy, in which a small sample of muscle from his calf was removed. By examining this

muscle sample, the doctor was able to find out what was actually happening inside the leg muscles and could determine what type of muscular dystrophy Bill had.

The doctor looked for a certain protein called dystrophin, which is present in muscle cells, but for those that have DMD , this protein is missing. MDA (2007)

Healing Approaches

Currently, there is no cure for DMD and the only medication that has improved muscle strength and function is the steroid prednisone. MarieB & Hoehn (2007). There is a promising technique called, myoblast transfer therapy, which involves injecting diseased muscle into a healthy myoblast cell, which fuses with the unhealthy cell. How this therapy works is that the normal gene provided via the healthy myoblast cell would allow the muscle fiber to produce dystrophin and allow the muscle to grow in a normal manner. This technique has been tested in mice, but human trials have not been proven as an effective treatment for DMD. MarieB & Hoehn (2007).

Miscellaneous

Bill's current condition is that his DMD is effecting his lungs and heart, in which he tires easily. This is due to his heart muscles and lungs muscles weakening. Like all muscles, the heart muscles and diaphragm also can be weakened by lack of dystrophin. Weakening of the heart is a condition called cardiomyopathy, (heart muscle weakness).Tremblay, J. (2006).

The weakness progresses until the heart fails.The diaphragm, which is a muscle that operates the lungs, may also weaken, making this muscle less effective during inhalation. Currently, Bill uses a cough assist device, which keeps the bronchial system free from secretions. This is very important, since anyone with a weakened respiratory system is also subject to more infections and difficulty in coughing. A simple cold can quickly progress to pneumonia. Metules, T. (2002).

Conclusion

While writing this paper, there were many learning lessons; however, what stands out and is the most meaningful lesson that I have learned from this paper is that people like Bill Watts have a strong desire to live life no matter what their circumstances are and have a strong will power to go through life carrying this burden without complaint. It is Bill's strength that people like me rely on. I will look back at Bill's determination to live when things in my life become challenging. I think of others like Bill, that have more burden and carry a heavier cross than I can ever imagine. I believe Bill's Catholic Faith, his devotion to Christ and His Church, is the main reason why Bill is a true survivor of DMD, since most pass away before their 21st birthday and Bill is 40 and he stated that his doctors cannot explain why he is still alive. It seems that some things can not be explained by science and medicine, since every human life is unique in its design and only God knows when it is time for us to return to his Heavenly Kingdom and depart this earth.

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