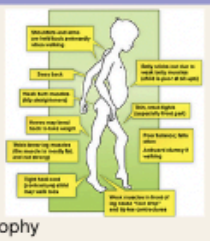
Hello, my name is Vincent and I am eager to be involved with the Rare Care program. With some background on research in malaria as one of my projects, I have developed a deep understanding and would be delighted to help patient care. Not only do I have some prior knowledge, but I believe that my abilities and skills are enhanced through my empathy and resilience. My experiences have taught me how important it is to resonate with patients and I am passionate about being able to make a difference. Finally, I hope I can bring these qualities to RareCare and make a lasting impact.

DUCHENNE MUSCULAR DYSTROPHY

By: Vincent Guo

What is Duchenne Musuclar Dustrophy?



Duchenne Muscular Dystrophy (DMD) is a genetic disorder caused by progressive muscle degeneration and weakness.

What causes DMD?

Duchenne muscular dystrophy is caused by a genetic problem in producing dystrophin, a protein that protects muscle fibers from breaking down when exposed to enzymes.

Current Treatment

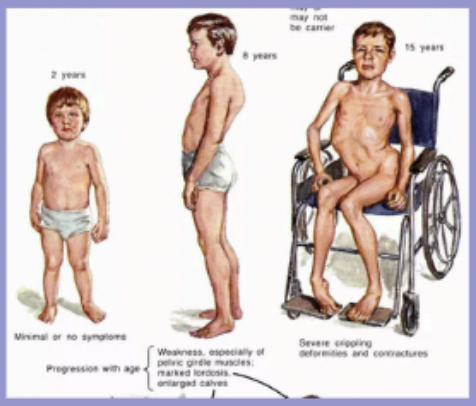
Corticosteroid medicine (steroids) has been shown to improve muscle strength and function for 6 months to 2 years, and slow down the process of muscle weakening.

Symptoms

- Muscle weakness
- Fatigue
- Difficulty walking
- Frequent falls
- Delayed growth.

Prognosis

- It is a progressive disease
- Most paitients need a wheelchair
- Short lifespan at around 20-30



Prevention?

There is no known prevention as it is a genetic disease

Duchenne Muscular Dystrophy (DMD) is a severe form of muscular dystrophy that primarily affects boys, though girls can be carriers and mildly affected. It is caused by mutations in the DMD gene, which encodes for dystrophin, a protein crucial for muscle fiber integrity. Without functional dystrophin, muscle cells are easily damaged, leading to progressive weakness and degeneration. There are three main stages to this disease, childhood, late childhood, and adults, each showing a different severity of the disease.

The first stage is childhood and the age ranges from 0-5. Duchenne is often diagnosed between the ages of 2 and 5, and abnormalities in sitting, walking, or talking, are noticed. Speech delay or the inability to keep up with peers will often be the first signs of this disorder. The treatment in this stage is aimed towards prevention of progression and maintaining strength and function of the muscles. Some safety precautions are to allow children to have resting periods. Children with Duchenne tire more quickly than peers, and while post children are good at self-limiting their activity so that they don't overwork, it is important to allow more rest periods so they do not injure themselves trying to "keep up."

The second stage is young childhood and the age ranges up to 12 years old. By about 12 years of age, most people with Duchenne are unable to walk and need to use a power wheelchair on a regular basis. At this age, the damages get detrimental and one may have more trouble with headaches, mental lapses, or difficulty concentrating or trouble staying awake during the day. Many experts recommend that people who are on steroids continue to take them when they stop walking. Sometimes doctors will change the dose of steroids after one stops walking. Continuing steroids can help keep the muscles of the upper body stronger, slow down scoliosis, and help keep the heart functioning properly.

The third stage is adulthood. The age ranges at most up to 30 years old. Here, it's almost the end of one's lifespan. If someone infected is taking steroids, it is imperative they are not stopped suddenly for any reason. This is risky for an adrenal crisis which is a life-threatening condition. Adults with Duchenne have more trouble using their hands and maintaining good posture. Weakness continues during the adult phase. There are also two problems that are common in the hearts of adults with Duchenne. The heart muscle may become weak and not pump blood properly. There are also problems with heart rate or rhythm. There is not much treatment or anything you can do at this point.

Most of the symptoms of the disease start noticing between the ages of 2–5. These might include frequent falls, difficulty performing running or jumping, and enlarged calf muscles. Subsequently, there is slowly progressive, pronounced floppiness beginning first in the legs and pelvis, proceeding to the arms, neck, face, and other areas. Most individuals by their adolescence with DMD show a requirement for a wheelchair. Moreover, DMD can be complicated, as in cardiomyopathy and respiratory difficulty, and can mostly be fatal in patients. Currently, there is no cure for DMD, but treatments aim to manage symptoms and improve quality of life. Corticosteroids, such as prednisone, are commonly used to slow muscle degeneration and improve strength. Physical therapy helps maintain muscle function and prevent contractures. Experimental treatments, including gene therapy and exon skipping, are showing promise in clinical trials.

As DMD is a genetic disorder, there is no prevention for it. Family genetic counseling, however, is still recommended for those families with a history of DMD so that individuals will

be aware of the risks and options. The prognosis for DMD is very poor; most of the patients die in their 20s or 30s due to respiratory or cardiac complications. Medical advancements regarding devices such as ventilators and improvement in cardiac care have increased the survivability years and have aided better living for many DMD sufferers.

I watched Ryan’s story and it was a moving video. While it also touched on the science side, there were also shots of his parents and the resilience that they had to go through when they found that their son was diagnosed with DMD. I was surprised to see that he was one of the patients that was chosen to do a special test called “exon skipping.” The results seemed quite positive and the result may be a worldwide contribution and could save the lives of many. The video also provides valuable information about DMD, explaining the genetic mutation that causes the disease and the lack of dystrophin protein, which is crucial for muscle strength and function. It discusses the various treatments and therapies available, such as physical therapy, corticosteroids, and emerging treatments like gene therapy and exon skipping. The family's proactive approach to exploring all possible treatment options underscores the importance of medical research and advancements in the field.