Duchenne muscular dystrophy essay

Burns – 2 Imagine being a parent and discovering that your child won’t be able to walk by the age of 12 and won’t live past the age of 25. (http://dystrophy. com/muscular-dystrophy/Types+of+Muscular+Dystrophies) This has become a reality for many parents as they find out that their child has a form of muscular dystrophy.

This particular type of muscular dystrophy is referred to as Duchenne Muscular Dystrophy, which is a genetically inherited disorder that is the most common and severe of all muscular dystrophies. It is found in 1 of every 3, 500 males and is characterized by progressive muscle wasting. It is caused by the lack of dystrophin, which is a protein found in the cell membrane of muscles. (http://www. answers.

com/topic/duchenne-muscular-dystrophy? cat= health) One early clinical sign of the disease is the child showing a late start in walking and sometimes they are referred to as a late bloomer. Usually when the child walks they have a waddling motion and sometimes they walk up on their toes. Usually, hypertrophy, or enlarging of the muscles, especially in the calves is noticed. However, the hypertrophy is actually caused by adipose (fat) tissue, which replaces the muscle tissue. Another clinical sign of DMD is small amounts of mental retardation or learning disabilities.

Although this is not apparent until the child is older, it is a common pattern that is found in children with DMD. There have been many studies done to determine what exactly causes the mental retardation. (http://en. wikipedia. org/wiki/Duchenne_muscular_dystrophy) Burns – 3Duchenne’s muscular

dystrophy is a recessive genetic disorder caused by a deficiency of the protein dystrophin, which is found in muscle cells.

This disorder is usually only found in males. It has very rarely been found in females because they have two X chromosomes and the disease is recessive. They pass it on to one half of their sons and to one half of their daughters who become carriers. In the possibility that the male with DMD reproduces, his sons would not be infected however his daughters would be 100% carriers of the disease. (http://www.

nlm. nih. ov/medlineplus/ency/article/000705. htm) Even though DMD is present from the initial stages of fetal development, physical signs are not present until 18 months to 4 years of age.

In addition to the clinical signs mentioned above, a child with DMD is unable to climb or pull itself up from the floor. Between the ages 3 and 7, a child with DMD may show signs of improvement, but that is due to natural growth and development. In school they start to fall behind in being able to keep up with their peers in physical activity. This sometimes leads to them being called clumsy or lazy. When the child is around 9 years, the disease starts to progress rapidly and by age 12, they can no longer walk on their own.

In the late stages, a significant loss of muscle tissue is noticeable. There may also be an increase of the curvature of the spine. (http://www. umm. edu/ency/article/000705.

htm) Burns – 4 When they are brought to the doctor, a series of tests are done to determine the problem. When a biopsy is done, the doctors find that

the muscle tissue has hypertrophied with adipose tissue. Shortly after the cells have become full of adipose tissue they die. Unlike other tissues of the body, muscle and nerve tissues cannot reproduce. An individual is born with a specific amount of muscle and nerve cells. Once they die, they cannot be regenerated.

This is bad news for people with DMD and they usually die around the age of 20 due to the diaphragm or heart muscle degenerating. There are several ways to test for Duchenne Muscular Dystrophy. The first is a blood test, called the serum creatine kinase test, looking for an increased amount of creatine kinase. Normally there is only a small amount of creatine kinase in the bloodstream. All muscles produce creatine kinase and release small amounts, however when the muscle cells tear from contracting, the creatine kinase leaks into the bloodstream. (http://www.

mdausa. org/Publications/Quest/q71ss-cktest. html) The second test that could be done is an electromyopathy test. This involves putting a small needle in the skin and recording the pattern of electricity.

When muscles contract, there is an electric current flowing through the tissue. (http://www. emedicinehealth.
com/electromyography_emg/article_em. htm) Burns – 5The third way to test for the disease is a muscle biopsy.

In this process a piece of muscle is removed and examined under the microscope. When the cells are seen as enlarged and filled with adipose tissue then the disease is prevalent. Another sign is when the muscle cells are already dead. (http://en.

wikipedia. org/wiki/Muscle_biopsy) The last way to test is direct DNA testing. While the child is still in the womb it can be tested for the disease by doing an amniocentesis. In this procedure, fluid and fetal cells are taken out from the amniotic layer that surrounds the fetus. Then the cells are checked for a defect or genetic mutation in the DNA.

(http://www. medgen. ubc. ca/robinsonlab/mosaic/clinical/prenatal/amnio. htm) At this time, there are many research studies being done to try and find a cure for Duchenne Muscular Dystrophy.

Unfortunately, a definite cure has not yet been found. Even though there is no cure for Duchenne Muscular Dystrophy, there are places people with the disease can go to get help. There are several Support Groups available to people with DMD. The Muscular Dystrophy Association or MDA is probably the largest support group. They offer financial and emotional support as well as a vast information site for all of the different types of muscular dystrophy. They fund research projects that are aimed at finding a cure for the disease and also publish their own bimonthly magazine called “ Quest.

” They can be found on the Internet at www. mdausa. org. Another support group is The Parent Project for Muscular Dystrophy Research. Like the MDA, they also support families with DMD and fund research projects.

(http://www. parentprojectmd. org/site/PageServer? pagename= nws_index)Duchenne Muscular Dystrophy is a severe form of muscular dystrophy that causes progressive muscle degeneration. Its main cause is the lack of the protein dystrophy that is necessary for the muscles to function properly. It is tough for the victims and their families because there

is no definite cure and they usually don’t live past the age of 20. Hopefully in the future, technology will enable researchers to be able to find a cure that will be a 100 percent effective when used.

Until that time comes, all DMD victims can do is wait and hope.