Duchenne muscular dystrophy essay examples

Science, Genetics

\n[toc title="Table of Contents"]\n

\n \t

1. \n \t
2. Clinical Presentation \n \t
3. Pathophysiology \n \t
4. Usual Age of Onset \n \t
5. Relevant cultural/ethnic/risk information \n \t
6. Genetic Issues in Duchenne Muscular Dystrophy \n \t
7. Diagnosis and Management of Duchene Muscular Dystrophy \n \t
8. Conclusion \n \t
9. References \n

\n[/toc]\n \n

This essay critically examines the nature of duchenne muscular dystrophy. The condition, affecting 1 out of 3, 600 boys, is an inheritable disorder arising due to mutation of the dystrophin gene. To a great extent, duchenne muscular dystrophy can be effectively managed by controlling the symptoms that lead to its development. However, currently, there is no cure for the condition, but with advances in medical technology there is hope for a complete control of the disease through gene therapy.

Clinical Presentation

Duchenne muscular dystrophy is a disease which causes weakening of muscle and leads to eventual death. The condition appears in the form of degenerated muscles around the legs and the pelvis region. Clinical diagnosis may reveal abnormal heart muscles, irregular rhythm of the heart,

wastage of muscles, and respiratory disorders (Kohler et al., 2009). Other symptoms include the inability to stand upright without aid, inability to walk upstairs and enlarged deltoid muscles.

Pathophysiology

With time, the muscles around the arms and the neck get severely affected. This inhibits the proper coordination of the motor functions, which affects running and walking activities. Consequently, the affected child falls frequently and may experience difficulties in climbing upstairs. By the age of twelve, the child may totally lose the ability to walk. Duchenne muscular dystrophy is also characterized by fatigue and failing of the cognitive abilities. As a result, learning difficulties arise. In some cases, failure of cognitive ability leads to mental retardation (Kohler et al., 2009).

Usual Age of Onset

Duchenne muscular dystrophy starts during infancy, but the symptoms are clearly visible by the age of five. The average life expectancy for affected individuals is around 25 years.

Relevant cultural/ethnic/risk information

Duchenne muscular dystrophy is an inheritable disease. Its symptoms are more visible in boys than in girls, although both sexes may have the mutation. This is because women are carriers of the mutant gene. Individuals with a family history of duchenne muscular dystrophy are at risk of inheriting the disease. However, it is also possible for mutations in the dystrophin gene to occur spontaneously.

Genetic Issues in Duchenne Muscular Dystrophy

Duchenne muscular dystrophy occurs due to mutation of the DMD gene (the gene is responsible for encoding the muscle protein dystrophin). The condition passes on to the next generation through recessive X chromosomes (Swaminathan et al., 2009). Males affected by the condition lack the capacity to make dystrophin in their protein muscles. Naturally, male offsprings have XY chromosomes. The X and Y chromosomes comes from the mother, and the father respectively. As a result, if the X-chromosome has DMD mutation, the male offspring will have DMD. On the contrary, females have two sets of the X-chromosome. This means that incase one set of the X-chromosomes cannot produce the dystrophin protein the other set acts as a backup copy, and can produce the protein. As a result, women can only be “ carriers”.

Most women with duchenne muscular dystrophy may be unaware of their condition since its symptoms are not clearly visible in females, unless they have a family history. However, recent research indicates that a small fraction of women affected by the condition show muscle weakness, which might be accompanied by heart disorders. Studies also indicate that carrier females have a 0. 5 chance of passing it to their children, with each pregnancy.

Male children born with mutant copy of the DMD gene have duchenne muscular dystrophy since they have a Y-chromosome, but lack back-up X chromosome. Affected males who go ahead to have children will sire carrier daughters, but none of the sons would be affected.

Diagnosis and Management of Duchene Muscular Dystrophy

Diagnostic process

In order to determine whether patients have duchenne muscular dystrophy, muscle biopsy needs to be done. The procedure aims to determine whether the individual has abnormal dystrophin levels in the muscles. The dystrophin protein can be seen after staining a sample using a particular dye. For an individual with sufficient quantities of dystrophin in the muscle, samples show a closely knit pattern of cells. However, for individuals with Duchenne, the samples do not show a closely knit pattern of cells due to lack of dystrophin. Sometimes, the biopsy test reveals some individuals with an intermediate amount of dystrophin protein. These individuals lie in the category of people with Becker muscular dystrophy.

Another diagnostic means is the use of genetic testing. Genetic testing involves examination of the body's genetic instructions on a blood sample. This helps to establish changes occurring in the DMD gene. There are various approaches to genetic testing such as sequencing and duplication. The methods can detect genetic changes causing the disease in close to 95% of the patients (Swaminathan et al., 2009). However, genetic testing is not a conclusive means to detecting duchenne muscular dystrophy. As a result, the muscle biopsy is the only reliable means of detecting the dystrophin protein level. In spite of this fact, a combination of techniques such as use of family history, clinical findings and muscle biopsy may be necessary in order in accurately diagnosing dystrophin protein.

Medial management

The most effective means to manage duchenne muscular dystrophy should be directed at the symptoms. This should start with a vigorous management of the heart disorder using anti-congestive medications. In some cases, it is necessary to carryout cardiac transplantation. This can go along with the use of other devices to counter respiratory complications. A steroid, prednisone, can be administered to improve strength in affected individuals.

Prognosis

However, it is possible for side effects to arise due to the use of prednisone. The side effects include behavioral changes, sudden weight increase and occurrence of high blood pressure. As a possible means to eliminating the side effects, medical practitioners recommend the use of Deflazacort. It is also vital to use physical therapy, which aids in facilitating movement and prevents contractures. After sometime, surgery may help in alleviating severe contractures and scoliosis. Research into the use of gene therapy in treating duchenne muscular dystrophy is still in the pipeline, and this might be a pleasant relief to the patients.

Conclusion

Duchenne muscular dystrophy is a medical condition which usually affects boys. Broadly speaking, it is an inheritable condition, although there are some cases of spontaneous mutation of the dystrophin gene for people without a familial history of the disease. Patients affected by the condition are unable to perform gross motor functions such as walking, running and

climbing stairs. Although the current medical technologies are unable to completely cure the condition, there is a great promise to control the disease in future- especially with the use of gene therapy.

References

Kohler, M., Clarenbach, C. F., Bahler, C., Brack, T., Russi, E. W., & Bloch, K. E. (2009). Disability and survival in Duchenne muscular dystrophy. Journal of neurology, neurosurgey & psychiatry, 80, 320-325.

Swaminathan, B., Shubha, G., Shubha, D., Murthy, A. R., Kumar, H. K., Shylashree, S., et al. (2009). Duchenne muscular dystrophy: A clinical, histopathological and genetic study at a neurology tertiary care center in southern India. Neourological society of India, 57 (6), 734-738.