Case Study no.2

Patient with Duchesne's Muscular Dystrophy

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Case Report

# Introduction

This paper analysis the case of Duchene’s Muscular Dystrophy. Duchenne is a progressive muscle disorder that are sources the harm of its purpose and so the pretentious ones turn out losing their individuality totally. The illness is caused by a transformation in the gene that codes for dystrophin, a protein that is critical to muscles. Its absence causes the muscle cells to be certainly impaired. Progressive muscle flaw leads to severe medicinal difficulties and children need a wheelchair around age 12 (Bach, et.al. 1997).

The mutation of the gene that is reasons Duchenne is conveyed, generally, from the mother to the child. Though, thirty-five percent of cases happen due to spontaneous change, and as the association of relatives of the affected people states. it can occur in any family, knows no borders and affects all cultures and races.

# FDA Regulation

The United States Food and Drug Administration (FDA) nowadays accepted the injection Exondys 51 (eteplirsen), the initial drug accepted to give patients suffering from Duchenne muscular dystrophy (DMD) . It is precisely designated for patients who have a deep-rooted change of the dystrophin gene responsible for skipping exon 51, which influences regarding thirteen percent of the people by DMD.

"Patients with a certain type of Duchenne muscular dystrophy will now have access to approved treatment for this devastating and rare disease. "In rare diseases, the formulation of new drugs is especially difficult due to the small number of people affected by each of these diseases and the lack of medical understanding of many disorders. Accelerated approval makes this medication available to patients with support in the initial data, but we look forward to learning more about its effectiveness through a clinical trial of ratification that the company must perform after approval. "

# Scientific Advancement

The early detection of complications associated with Duchenne muscular dystrophy (DMD) allows the identification of patients with this disease to channel them to appropriate treatment. This will depend on the diagnosis and timely follow-up, for which the participation of a multidisciplinary group of high specialty is necessary and needs the collaboration of the patient and his family. In addition, they emphasize the importance of the early integration of the clinical-molecular diagnosis to be able to carry out the appropriate interventions and thus avoid the rapid progress of the disease.

# Role of Family

A Duchenne diagnosis will alter the whole family dynamics since all the attention will rest on the child with the diagnosis. However, parents, family and friends should not forget that other children are also struggling with the changes around them.

Supposedly, grandparents must have a more accessible time. They have already offered their support to their own children and now they want to enjoy their grandchildren, pamper them and send them home with their parents. With a diagnosis of muscular dystrophy everything changes, now there are also concerns. They continue with their beloved grandson, perhaps they adore him even more. It is a very difficult position. Grandparents have a unique and challenging role to play in a family with Duchenne. Once again it is a role as educator, guide, supporter, hope and confidence in own family and others. The communication is the key. Whether as a family member or as a friend, they can help to ensure that parents communicate with each other, and that their other healthy siblings continue to get time and individual attention

# Description of Disease

Duchenne muscular dystrophy (DMD) is a progressive inherited disease that has inheritance X-linked recessive disease. half of the male members of the family, and the of female members are carriers of asymptomatic. It affects approximately 1 in 3,500 boys born alive. The abnormal gene is located in the short arm of X chromosome, Xp21 locus, subband Xp212. The gene under normal conditions is responsible by the production of a protein called dystrophin, located in the sarcolemma of muscle fibers 1-4 . Clinical manifestations usually begin childhood, usually in the first three years of life life. Functional changes begin with the in- muscular weakness, which occurs gradually and in an ascending, symmetrical and bilateral manner, starting pelvic girdle and lower limbs, progressing for musculature of the trunk and for the musculature res- responsible for the support of the biped, waist scapular, upper limbs, neck and muscles respiratory (Bonilla, et.al. 1988).

Muscle weakness becomes evident around five years of age, when children have initial symptoms, such as difficulty in walking, jumping and running, as well as frequent falls. The muscle strength both extensor of the knee and hip are not sufficient to allow the extension of the trunk when the patient rises from the soil, triggering the Gowers signal 1,2,4 . As the disease evolves the weakness of The gluteus medius and minimum gluteus muscles result in pelvis when the child stands upright; since, with the progression of the disease, this inclination is even more disturbed, assuming a typical aspect due to excessive movement in the pelvic girdle, the called myopathy or anserine gait. The patient loses the ability to wander, and then the wheelchair, approximately from 10 to 13 years of age.

# Laboratory Testing

## Observation:

Parents or teachers are often the first to notice the first symptoms of Duchenne, such as speech delay, enflamed calf physiques, and the incapability to continue with their peers.

## Genetic tests:

There are reliable tests to help doctors diagnose a child with Duchenne. Some of the tests are Creatine kinase and Genetic tests.

The diagnosis is suspected depending on the characteristic clinical signs, the age of onset of the disease and family history suggesting an X-linked recessive mode of inheritance. Myopathic changes are visible on electromyography (potentials of motor units increase rapidly, have a short duration and low amplitude) and muscle biopsy (necrosis and a noticeable change in the size of muscle fibers not separated from motor units). Creatine kinase levels are exceeded 100 times from the norm (Finder, et.al. 2004).

# Causes of the Disorder

Duchenne muscular dystrophy is produced through a defective gene for dystrophin (a protein in the muscles). It usually happens in people by families by no identified past of this condition. Due to the means the disease is congenital, men are pretentious and not women. The children of women transporters of the illness (women with a defective chromosome but asymptomatic) apiece have a 50% coincidental of having the disease and the daughters each have fifty percent chance of being carriers (Mendell, et.al. 2010).

This disorder usually occurs at the age of 2–3 years. Weakness affects the proximal muscles, usually the lower limbs first. Children often walk on their fingers, have a walk, waddle and lordosis. It is difficult for such children to run, jump, climb stairs and get up from the floor. They often fall and get broken arms or legs (approximately in 20% of patients). There is a stable progression of weakness, and almost all children develop flexion contractures of the limbs and scoliosis. Reliable pseudo hypertrophy develops (replacement of individual enlarged muscle groups with fatty or fibrous tissue, especially at the ankles). Most children are confined to a wheelchair under the age of 12 years and die of respiratory complications by the age of 20 (Nigro, et.al. 1990).

Consequences of cardiac muscle involvement include dilated cardiomyopathy, conduction abnormalities, and arrhythmias. Such complications occur in about a third of patients by the age of 14 and in all patients over the age of 18; however, since these patients are not able to exercise physical activity, heart damage is usually asymptomatic until the late stage of the disease. About one-third of them have mild non-progressive dementia, which affects verbal ability more than productivity.

# Conclusion

There is no known treatment for Duchenne muscular dystrophy and the goal of dealing is to control the symptoms to optimize the quality of life. A child diagnosed with Duchenne will require interventions at multiple levels, and the child's needs will change over time. Steroids can decrease the loss of muscle strength. The child can start taking them when diagnosed or when muscle strength begins to decline.

Several medications can help with cardiac activity, such as angiotensin-converting enzyme inhibitors, beta-blockers, and diuretics. It is necessary to evaluate and treat speech delay and language problems through a speech therapist. Exercises for muscles included in speech and articulation help are appropriate and necessary for both males who have Duchenne with difficulties in this area and for older individuals who have impaired oral muscle strength and / or incomprehensibility of difficult speech.

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