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February 5, 2013
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"Pompe Disease, A Rare but Fatal Disorder"

Pompe Disease is a rare genetic disorder that can affect both males and females. The disease causes many physical symptoms and can lead to death. If detected early, affected individuals can undergo treatment. New treatments are currently being researched.

Normally people have a gene that makes an enzyme called acid alpha-glucosidase (1). This enzyme is responsible for breaking down a complex sugar in the body called glycogen (2). Pompe disease is caused by a mutation to this gene (1, 2). This gene is inherited usually by both parents. Both parents would carry the autosomal recessive mutation. If both parents pass on this recessive gene their child will have Pompe disease (2). This is a rare mutation that only affects 1 in every 40,000 births (1). The mutation is in the GAA gene (2). Individuals with this mutation do not produce this vital enzyme. Therefore, sugar in the body builds up and does not get broken down properly. The built up sugar hinders the function of the person's muscles and tissues.

The symptoms of this disease are widespread. Babies born with the disease have to overcome many obstacles. Some of these include, having difficulty to eat and therefore low weight gain. These infants also generally have difficulty breathing. As a result, many pompe babies never make it past a year old (1). If an affected child does make it past their first birthday, they will continue to be faced with other symptoms. Some of these symptoms include abnormal brain function, liver enlargement, scoliosis, tongue enlargement, problems with joints and muscle development and heart problems (4). Overall, the muscles and tissues in the diseased body continue to deteriorate. This deterioration affects all of the major muscular functions in the body.

This disease can be easily detected by one of many possible tests that can be done. A tissue sample of the suspected individual can be tested for the GAA mutation, or the amount of sugar can be measured in this tissue sample (4). A positive result for the mutation and a high amount of glycogen in the tissue would point to the individual as having Pompe Disease. Prenatal testing can be done in cases where parents already have an affected child (5). There is also a test that can be given that will look for the acid alpha-glucosidase enzyme in the person's body. The enzyme is not found in people that have Pompe (4).

Research is currently being conducted on this disease. The National Institute of Neurological Disorders and Stroke (NINDS) is an organization that provides grants of money to research. Research focuses on finding a cure, or works on improving overall treatment of the disease (1). At Emory University, the Lysosomal Storage Disease Center in the Department of Human Genetics is currently caring for patients with Pompe. Along with care, the department is conducting research trials to try and increase the affectivity of detecting Pompe in newborns (5). MDA is an organization that conducts research on many muscular diseases and Pompe is one of them (3). Duke University is conducting many studies. Some of these studies include trying to find less invasive ways of surveying the disease, they are researching ways to increase respiratory and immune system function and trying to find ways in which diseased individuals can exercise (4). Overall, research seems to be more focused on treatment rather than cure.

Since no cure has been discovered, Pompe Disease is still proved to be fatal in some individuals. The Disease is rare, but can be inherited. Inheriting the disease will lead to the

individual having many disabling symptoms, especially in cases, where the individual is not treated.

Works Cited:

1. National Institute of Neurological Disorders. http://www.ninds.nih.gov/disorders/pompe/pompe.htm
2. Genetics Home Reference. http://ghr.nlm.nih.gov/condition/pompe-disease
3. Metabolic Diseases of Muscle. http://mda.org/disease/metabolic-diseases-of-muscle
4. Dukes Children's Hospital. http://www.dukechildrens.org/services/medical_genetics/pompe
5. Emory University. http://genetics.emory.edu/clinical/?assetID=1281