Homework One: Medical Disorders: Are they Genetic or Environmental?

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1.)What is it?

The U.S. National Library of Medicine's 2020 article "Maple syrup urine disease", Maple Syrup Urine Disease is a disorder that is inherited where the human body is not able to process amino acids properly. The inherited disorder has gotten its name from the sweet odor that comes from the affected infants' urine. There are various forms of this disorder that would become apparent later in infancy or childhood. In essence, the variant forms of this disorder are usually milder, yet it causes delayed development and other health problems for the individual if it is not treated (U.S. National Library of Medicine, 2020). Other names for Maple Syrup Urine Disease would be MSUD, ketoacidemia, branch-chain ketoaciduria, branched-chain alpha-keto aid dehydrogenase deficiency, and BCKD deficiency (U.S. National Library of Medicine, 2020).

2.)How do people get it (Genetic.... What genes/chromosomes? Is it Environmental?)

Individuals who get this inherited disorder is due to the copies of the gene in each cell that has mutations. According to the U.S. National Library of Medicine's 2020 article "Maple syrup urine disease", the parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition (U.S. National Library of Medicine, 2020). In the U.S. National Library of Medicine's article "Newborn Screening ACT Sheet [Increased Leucine] Maple Syrup (Urine) Disease", it stayed that the leucine, isoleucine, and valine (branched chain amino acids) cannot be metabolized further than their α-ketoacid derivatives. The amino acids and organic acids accumulate and produce severe toxicity (American College of Medical Genetics, 2012).

3.)What are the symptoms (Physical & cognitive if applicable)?

The physical symptoms of Maple Syrup Urine Disease are lethargy, weight loss, poor appetite, irregular sleeping patterns, a maple sugar odor in urine, sweat, and the earwax, feeling irritable, a high pitched cry, and having episodes of both hypotonia (the muscles are limp) and hypertonia (the muscles are rigid). In Medline Plus' article of Maple Syrup Urine Disease, it stated that other symptoms of this disorder include lethargy, seizures, and vomiting. There are possible complications from Maple Syrup Urine Disease that can lead to mental disability, neurological damage, coma, and death (Medline Plus, 2019).

4.)How do doctors diagnosis it (test/labs used)?

The doctors' diagnosis of Maple Syrup Urine Disease by doing genetic testing, plasma amino acid test, and urine organic acid test. When doing these tests on the individual, the doctors check for signs of both excessive acid in the blodod and ketosis. The Plasma Amino Acid Test is done on infants to check the amount of amino acids in the blood for the Maple Syrup Urine Disease (Medline Plus, 2019). The Urine Organic Acid Test is based on collecting a clean catch urine sample (Medline Plus, 2019). In genetic testing, the autosomal recessive condition must be checked in the individual since both parents of the person carry one copy of the mutated gene.

5.)What are the treatment options?

The treatment options for Maple Syrup Urine Disease are having the individual get involved in a protein free diet. When having this diet, sugars, fats, and fluids aredone through an IV, and dialysis is done through the individual's belly or vein in order to reduce the amount of abnormal substances in the bloodstream (Medline Plus, 2019). For long term treatment upon infants, their diet includes a formula with low levels of the amino acids of valine, isoleucine, and

leucine. People with this Maple Syrup Urine Diesease must have a diet with low amount of amino acids (Medline Plus, 2019).

References

American College of Medical Genetics ACT SHEET. (2012). Newborn Screening ACT Sheet [Increased Leucine] Maple Syrup (Urine) Disease. American College of Medical Genetics and Genomics. Retrieved August 20, 2020, from https://www.ncbi.nlm.nih.gov/books/NBK55827/bin/Leucine.pdf

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