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Commentary

Overview on Causes of Phenylketonuria Related Diseases and their Symptoms

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Introduction

Phenylketonuria is a genetic disorder that is passed down from one's parents. It is caused by PAH gene mutations, which result in low levels of the enzyme phenylalanine hydroxylase. As a result, dietary phenylalanine levels rise to potentially toxic levels. It is autosomal recessive, which means that for the condition to manifest, both copies of the gene must be mutated. Classic PKU and variant PKU are the two main types, depending on whether any enzyme function remains. Those who have only one copy of a mutated gene do not typically exhibit symptoms. Many countries have disease screening programmes for newborns. Treatment consists of a diet low in phenylalanine-containing foods as well as special supplements. Babies should be given a special formula that contains a small amount of breast milk. The diet should begin as soon as possible after birth and be followed for the rest of one's life. People who are diagnosed early and follow a strict diet can have normal health and life expectancy. Blood tests are used to monitor effectiveness on a regular basis. Sapropterin dihydrochloride, a medication, may be beneficial in some cases. The age at which symptoms of a disease first appear is referred to as the age of onset. The age of onset varies between diseases and may be used by a doctor to make a diagnosis.

Description

Symptoms of some diseases may appear in a single or multiple age groups. Symptoms of other diseases can appear at any time during a person's life. One in every 12,000 babies is born with phenylketonuria. Both men and women are affected equally. Ivar Asbjrn Flling discovered the disease in 1934, and the importance of diet was determined in 1935. Gene therapy, while promising, requires much more research as of 2014. PKU is treated by limiting protein intake to limit phenylalanine intake. If you or your child has moderate to severe PKU, your diet will need to be modified to include special formulas and meals. These changes include eating a low-phenylalanine diet and getting enough protein. It's also critical to avoid the artificial sweetener aspartame, which raises phenylalanine levels in the body. A dietitian who is familiar with PKU can assist you in developing a diet. This diet must be followed for the rest of your life. You will need frequent blood tests to monitor your phenylalanine levels as part of your treatment. If you have PKU and are pregnant, your doctor and a dietitian will work together to develop a diet plan that will ensure you get proper nutrition while also lowering your baby's risk of complications. Some PKU patients benefit from the medication sapropterin dihydrochloride (Kuvan®), which aids in the breakdown of phenylalanine.

PKU is inherited in an autosomal recessive manner. When a child inherits an abnormal gene from both parents, he or she develops a recessive genetic disorder. If a person receives one normal and one abnormal gene copy, they will be a carrier for the condition but will not experience symptoms. With each pregnancy, there is a 25% chance that two carrier parents will both pass the abnormal gene and have an affected child. Males and females are both at risk. There are over 300 different changes (mutations) in the PKU gene. Because different mutations result in varying degrees of PAH enzyme activity and thus varying degrees of phenylalanine elevation in blood, each child's diet must be tailored to their specific needs. Affected Populations reported incidence of PKU from newborn screening programmes in the United States ranges from one in 13,500 to one in 19,000 newborns. PKU affects

people of all ethnic backgrounds, though it is more common in African Americans and Jews of Ashkenazi ancestry.

Conclusion

The goal of PKU treatment is to keep plasma phenylalanine levels between 120-360 umol/L (2-6 mg/dL). This is usually accomplished through a well-planned and monitored diet. Because phenylalanine is an essential amino acid, it must be limited in the child's diet with caution. A well-balanced diet can help to prevent intellectual disability as well as neurological, behavioural, and dermatological issues. If treatment is not initiated at a young age, some degree of intellectual disability can be expected. However, some late-treated children have performed admirably. Children with PKU who are treated with a low phenylalanine diet before the age of three months do well, with an IQ in the normal range, according to studies. When people with PKU stop controlling their phenylalanine intake, neurological changes usually occur. IQ levels may fall. Difficulties in school, behavioural problems, mood changes, poor visual-motor coordination, poor memory, poor problem-solving skills, fatigue, tremors, poor concentration, and depression are some of the other issues that may arise and become severe once dietary regulation is discontinued.

Acknowledgment

None

Conflict of Interest

None