Commentary
Volume 11:10, 2022

Journal of Metabolic Syndrome

ISSN: 2167-0943 Open Access

Insights on Phenylalanine Hydroxylase Deficiency Disease

John Mitchell*

Department of Human Genetics and Pediatrics, University of Helsinki, Finland

Introduction

Phenylketonuria, also known as PKU, is a rare genetic condition that results in the buildup of the amino acid phenylalanine in the body. The gene that makes the enzyme needed to break down phenylalanine has a mutation that leads to PKU. When a person with PKU consumes protein-containing foods or aspartame, an artificial sweetener, a hazardous accumulation can occur because this individual lacks the enzyme necessary to metabolise phenylalanine. In the long run, this may result in severe health problems. For the remainder of their lives, PKU infants, kids, and adults must consume a diet low in phenylalanine, which is predominantly found in foods containing protein. Babies are screened for PKU soon after birth in the US and many other nations [1].

Description

PKU patients who get pregnant run the risk of having maternal PKU, a distinct type of the disease. Blood phenylalanine levels may increase if women do not follow the particular PKU diet before and during pregnancy, harming the developing foetus or leading to a miscarriage. If they do not follow the PKU diet, even women with less severe cases of PKU risk endangering their unborn infants. Babies born to moms with high phenylalanine levels hardly ever inherit PKU. However, they may have detrimental effects if the mother's blood phenylalanine level is high during pregnancy.

Genetic mutations, which can be minor, moderate, or severe, are what lead to PKU. A person with PKU will lack or have insufficient amounts of the enzyme needed to break down the amino acid phenylalanine as a result of this faulty gene. A dangerous buildup of phenylalanine can happen when a person with PKU consumes protein-rich foods like milk, cheese, nuts, or meat, as well as grains like bread and pasta, as well as when they take aspartame, an artificial sweetener. In the brain, this phenylalanine accumulation damages nerve cells. [2].

Both the mother and the father must have the faulty gene and pass it on to the child in order for the child to inherit PKU. This mode of inheritance is called autosomal recessive. A parent may be a carrier, meaning they may carry the PKU-causing gene aberration but not the disorder. If just one parent carries the faulty gene, there is no danger of transferring PKU to the child; nonetheless, the child may be a carrier. Two parents who have PKU but are ignorant of it are most likely to pass it on to their children. PKU is brought on by both parents carrying a faulty gene. Both parents must be affected by the condition for their child to have it. [3,4].

PKU can only be inherited if both the mother and the father have the faulty gene and pass it on to the offspring. Autosomal recessive refers to this mode of inheritance. A parent can be a carrier, meaning they have the PKU-causing gene flaw but do not experience the illness. If just one parent carries the problematic gene, there is little danger of PKU being passed on to the child, although the youngster might be a carrier. Two parents who are carriers of the illness but are unaware of it are most frequently the source of PKU in offspring. PKU is brought on by the presence of a faulty gene in both parents. Both parents must be affected for the condition to manifest in their child. [5].

Conclusion

For the rest of your life, you should take a PKU formula, a specialised dietary supplement, to make sure you obtain enough nutrients and vital protein (free of phenylalanine) for growth and general health. With PKU, a safe phenylalanine intake varies from person to person and is subject to fluctuate over time. The general rule is to limit phenylalanine intake to that which is necessary for healthy bodily function and growth. Your doctor can establish a healthy quantity by routinely checking your blood phenylalanine levels, growth charts, and food records. regular blood tests to monitor phenylalanine levels as they change over time, especially during pregnancy and childhood development spurts.

Acknowledgement

None.

Conflict of Interest

None.

References

1. Mayer P., J.L. Pépin, G. Bettega and D. Veale, et al. "Relationship between body mass index, age and upper airway measurements in snorers and sleep apnoea patients." *Eur Respir J* 9 (1996): 1801-1809.
2. Ward, Richard A., Bärbel Schmidt, Jeannine Hullin and Günther F. Hillebrand, et al. "A comparison of on-line hemodiafiltration and high-flux hemodialysis: A prospective clinical study." *J Am Soc Nephrol* 11 (2000): 2344-2350.
3. Semenza, Gregg L., and Reed E. Pyeritz. "Respiratory complications of mucopolysaccharide storage disorders." *Med* 67 (1988): 209-219.
4. Delanaye, Pierre, Bernard E. Dubois, François Jouret and Jean-Marie Krzesinski, et al. "Parathormone and bone-specific alkaline phosphatase for the follow-up of bone turnover in hemodialysis patients: Is it so simple?." *Clin Chim Acta* 417 (2013): 35-38.
5. Phan, T. C. A., Jiake Xu and M. H. Zheng. "Interaction between osteoblast and osteoclast: Impact in bone disease." *Histol Histopathol* 19 (2004).

***Address for Correspondence:** John Mitchell, Department of Human Genetics and Pediatrics, University of Helsinki, Finland, E-mail: JohnMitchell22@gmail.com*

Received: 03 October, 2022, Manuscript No. jms-23-87763; **Editor Assigned:** 05 October, 2022, PreQC No. P-87763; **Reviewed:** 17 October, 2022, QC No. Q-87763; **Revised:** 24 October, 2022, Manuscript No. R-87763; **Published:** 31 October, 2022, DOI: 10.37421/ 2167-0943.2022.11.303

How to cite this article: Mitchell, John. "Insights on Phenylalanine Hydroxylase Deficiency Disease." J Metabolic Synd 11 (2022): 303.