PHENYLKETONURIA

Its an inherited birth defect that causes amino acids known by phenylalanine to build up in the body.

Phenylalanine is an essential amino acid, which is mainly found in high protein foods such as eggs, almonds, beans. Its main purpose is to improve learning ability and memory. It is also involved in enhancing alertness and the mood of an individual.

It is also used in the management of certain conditions such as certain types of depressions, schizophrenia, Parkinson disease, in the management of obesity where it is administered to reduce a person’s appetite hence reducing the amount of food intake.

Phenylketonuria is caused by phenylalanine hydroxylase gene. The gene involved in manufacturing an enzyme needed to breakdown the amino acid phenylalanine to. This enzyme is known as phenylalanine hydrolase. Defect in the gene causes failure of production of phenylalanine hydrolase, where the amino acid phenylalanine cannot be broken-down.

Failure of breaking down of the phenylalanine causes a buildup which in turn brings about toxicity. Toxicity can be associated with intellectual problems.

Some of the signs and symptoms include: skin rash similar to that of eczema, seizures, microcephally (abnormally small head), hyperactivity.

Phenylketonuria can be subdivided into two types: classic and less severe form.

Classic from. In this form the enzyme needed in the breakdown is usually missing or in reduced amounts. This in turn causes a rise in the levels of phenylalanine. It is characterized by more severity in symptoms such as, mental retardation, organ damage,

Less severe form (mild/moderate). In this form the enzyme is present and functional but there are minimal levels of phenylalanine. There are less severe symptoms and less chances of having a brain damage occurring.

Complications that may arise from the above condition include irreversible brain damage, neurological problems such as seizures, tremors delayed developments in children.

Risk factors include a family history of the same condition.

A diagnosis of phenylketonuria can be made by screening the baby’s blood. That is after birth a sample if the baby’s blood is collected and taken to the laboratory for further studies.

Treatment modalities: dietary modification; intake of low phenylalanine foods, and avoid foods such as eggs, milk cheese beans and peas.

Sapropterin is a drug that has been identified in the management of phenylketonuria. A combination of the drug and proper diet improves the well being of people involved.

Special nutritional supplements that does not contain phenylalanine but contains essential nutrients that are needed for growth and development.

Feeding in babies and toddlers a phenylalanine free diet is made and given to the babies for feeding since breast milk contains phenylalanine.

Staying informed and offering general knowledge about phenylketonuria, regular observance of the types of food being eaten, regular follow up from medical professionals.

VERY LONG CHAIN Acyl-COA

Refers to a metabolic disorder that presents when the body in unable to convert fatty acids into energy during times in which energy is more essential in the body like in periods such as fasting, periods or starvation.

A long chain Acyl-COA is an enzyme that is involved in the process of activating fatty acids in order for them to undergo metabolism (Co-enzyme A). It attaches to a long chain of fatty acids hence forming Acyl-COA.

Its main involvement in lipid metabolism, synthesis of complex lipids and energy production.

Very long chain Acyl-COA is linked to several conditions which include; X-linked adrenoleukodystrophy, it is a chromosome X linked disease that is as a result of buildup of fatty acids due to failure of beta oxidation of fatty acids. This results to formation of a very long chain of fatty acids that accumulates in the brain. It is characterized by; neurological deficits, adrenal insufficiency due to accumulation of very long chain Acyl-COA.

Zellwegers spectrum disorders: also known as cerebrohepatorenal, it is a condition associated with either reduction or complete loss of peroxisomes. Peroxisomes are necessary in carrying out oxidative reactions in the cells.

Refsum disease, refers to accumulation of phytanic acid. Caused by a lack of phytanoyl-CoA(Co enzyme A) hydroxylase. It is used in breakdown of phytanic acid. It is characterized by neuropathy, loss of hearing, diminished muscle coordination.

It exists in various forms according to the stage of development; early onset, during infancy, characterized by low blood sugars, lethargy. Childhood onset, characterized by enlarged liver, and low blood glucose levels. In adults symptoms can be triggered by certain factors which include; fasting, illness, exercise, exposure to variant temperatures.

Diagnosis of this disease is made during infancy, where the baby’s blood is screened to check for abnormally elevated levels. A heel prick is done and a sample collected which aids in identifying levels in blood.

Treatment of this condition includes nutritional support; feeding the child at intervals to avoid periods of fasting or starvation which brings about low blood sugars which might complicate further, low fat diet in order to reduce the metabolism rate of fatty foods, close follow up with a professional, reduction of physical activity.

Carnitine is a drug that is recommended and is usually used for turning fats into energy in the body. Carnitine is a chemical that is produced by the body, but in this case the baby’s body is not capable of producing enough amounts needed to break down fats into energy.

Vitamin B2 can also be given as a supplement.

Early detection and diagnosis is essential for a better treatment and