Phenylketonuria

Phenylketonuria, also called PKU, is an inherited genetic condition where the body cannot use an amino acid, called phenylalanine (PHE). PHE cannot be broken down by the body, so it builds up in the bloodstream, affecting development. This buildup of PHE can slow development if not properly treated. Without treatment, PKU results in menta retardation within the first year after birth.

PKU is caused by a recessive gene, so a patient with PKU has inherited mutated genes from both parents. Since the condition is recessive, children of one PKU parent runs a better than 50-50 chance of not inheriting the condition themselves. Routine blood test after birth will reveal if a child suffers from PKU. The syndrome occurs in approximately 1 in 10,000 live births.

Effects of Phenylketonuria

Phenylketonuria is particularly detrimental in the early years of development and the greatest period of growth. Without treatment of the syndrome, the PHE buildup will cause progressive brain damage. To prevent significant damage, treatment must begin within the first few weeks after birth.

Patients with PKU must follow a low-PHE diet. Researchers disagree on how long patients must follow this diet. Patients must adhere to the diet at least through developmental years, and women who wish to become pregnant should follow the diet. Women with PKU who abandon the special diet put their children at high risk for mental retardation and other disorders if they choose to bear children. Some doctors, however, suggest the low-PHE diet should be followed throughout life.

Treatment

PKU is treated through diet low in PHE but high in supplementary amino acids which will aid development in the absence of PHE. Patients with PKU must consult a dietician to develop a proper tailored treatment program. The diet can be modified for a variety o reasons, including illness, development stages, or eating habits.

Babies with PKU must be fed a special formula in replacement or supplement of breas milk which contains PHE. The PKU diet prohibits proteins such as those found in milk and meats. Flour and pastas must be made with special ingredients. Many PKU foods are not easily available. The diet can be difficult to maintain. Patients may have to order food through distribution centers for people with medical conditions.

What teachers should know

Classroom teachers should be aware of any students with PKU in order to help monitor the child's condition and observe the student's diet (particularly during field trips or other non-routine activities).

Patients with PKU should avoid diet food or drinks since some sweeteners including NutraSweet are made from PHE. Students may need several days notice before a die alteration (such as a class party) so their diet may be adjusted in advance to allow participation.

There are signals of PHE imbalance to watch for. Skin rashes or diarrhea can indicate too little PHE in the blood. Children should also be especially closely monitored during illness. Infections can cause a normal increase in PHE levels, and diet may need to be adjusted accordingly.

Sources of informatio

Phenylketonuria Newsletter
http://www.pkunews.org/

Phenylketonuria Information
http://www.mcgill.ca/pahdb/handout/handout.htm

Phenylketonuria Technical Informati
http://www3.ncbi.nlm.nih.gov/htbin-post/Omim/dispmim?261600