

Disorders of amino acid metabolism

Metabolism is the process your body uses to make energy from the food you eat. Food is made up of proteins, carbohydrates, and fats. Your digestive system breaks the food parts down into sugars and acids, your body's fuel. Your body can use this fuel right away, or it can store the energy in your body. If you have a metabolic disorder, something goes wrong with this process.

One group of these disorders is amino acid metabolism disorders. They include phenylketonuria (PKU) and maple syrup urine disease. Amino acids are "building blocks" that join together to form proteins. If you have one of these disorders, your body may have trouble breaking down certain amino acids. Or there may be a problem getting the amino acids into your cells. These problems cause a buildup of harmful substances in your body. That can lead to serious, sometimes life-threatening, health problems. These disorders are usually inherited. A baby who is born with one may not have any symptoms right away. Because the disorders can be so serious, early diagnosis and treatment are critical. Newborn babies get screened for many of them, using blood tests.

Treatments may include special diets, medicines, and supplements. Some babies may also need additional treatments if there are complications. Classic (hepatorenal or type I) tyrosinemia is caused by a deficiency of fumarylacetoacetate hydrolase (FAH), the last enzyme in tyrosine catabolism. Features of classic tyrosinemia include severe liver disease, unsatisfactory weight gain, peripheral

nerve disease, and kidney defects. Approximately 40 percent of persons with the disorder develop liver cancer by the age of 5 if untreated. Treatment with 2-(2-nitro-4-trifluoromethylbenzoyl)-1,3-cyclohexanedione (NTBC), a potent inhibitor of the tyrosine catabolic pathway, prevents the production of toxic metabolites. Although this leads to improvement of liver, kidney, and neurological symptoms, the occurrence of liver cancer may not be prevented. Liver transplantation may be required for severe liver disease or if cancer develops. A benign, transient neonatal form of tyrosinemia, responsive to protein restriction and vitamin C therapy, also exists.

Non-ketotic hyperglycinemia is characterized by seizures, low muscle tone, hiccups, breath holding, and severe developmental impairment. It is caused by elevated levels of the neurotransmitter glycine in the central nervous system, which in turn are caused by a defect in the enzyme system responsible for cleaving the amino acid glycine. Drugs that block the action of glycine (e.g., dextromethorphan), a low-protein diet, and glycine-scavenging medications (e.g., sodium benzoate) may ease symptoms, but there is no cure for this severe condition.