Inborn Errors of Metabolism

What Are Inborn Errors of Metabolism?
Inborn errors of metabolism are rare genetic disorders in which the body is unable to metabolize, or break down, certain nutrients. These disorders may also be referred to as inherited metabolic diseases, congenital metabolic diseases, or hereditary metabolic disorders.

Individuals with these conditions must follow a strict diet because unmetabolized nutrients will build up in their bodies and become toxic. This can result in serious complications, including mental retardation, organ failure, or death.

There are many different forms of inborn errors of metabolism that may affect children; just a few examples of these are discussed below.

Phenylketonuria (PKU)
A deficiency of the enzyme that processes the amino acid phenylalanine.
• Health Risks–Elevated levels of phenylalanine in the blood and body tissues can lead to mental retardation, delayed speech, seizures, eczema, and behavior abnormalities.
• Dietary Treatment–A strict low-protein diet; no meat, fish, milk, cheese, or eggs. Some vegetables, fruits, and grains may be restricted depending on the individual’s level of phenylalanine tolerance. Special protein formulas are required daily to provide essential nutrients. Aspartame-containing foods, such as diet drinks, should be avoided since aspartame contains phenylalanine.

Galactosemia
A deficiency of the enzyme that breaks down galactose, a sugar found in milk and dairy products.
• Health Risks–Elevated levels of galactose in the blood can lead to liver, brain, eye, and kidney damage.
• Dietary Treatment –No milk or dairy products should be consumed. Because milk is found in many processed foods, it is important to check food labels for ingredients such as milk, butter, cheese, nonfat dry milk solids, lactose, casein, whey, whey solids, sodium caseinate, calcium caseinate, and lactalbumin.

Hereditary Fructose Intolerance
The body does not produce the enzyme to metabolize fructose, a sugar found in many foods and sweeteners.
• Health Risks–Elevated levels of fructose in the body will lead to low blood sugar, liver damage, or liver failure.
• Dietary Treatment–Fructose should be eliminated from the diet; no fruit, honey, table sugar, or commercial sweeteners, such as high-fructose corn syrup. It is important to check food labels because sugar and high-fructose corn syrup are common ingredients in many processed foods and beverages.
Inborn Errors of Metabolism, continued

**Maple Syrup Urine Disease**

The body is unable to metabolize the amino acids leucine, isoleucine, and valine. (This inborn error of metabolism is named for the distinct odor of the affected individual’s urine.)

- Health Risks—Build-up of leucine, isoleucine, and valine can lead to neurological damage and death.
- Dietary Treatment—The diet prescribed is similar to that for PKU; limitation of protein sources such as meat, fish, milk, cheese, or eggs. Special protein supplementation may be required to meet nutrient needs.

**Advice for School Nutrition Assistants**

Children with these and other inborn errors of metabolism may require special accommodations in the cafeteria. Careful food selection and preparation by school nutrition assistants will help ensure that these children can safely participate in the school nutrition program. Some tips to follow include:

- Maintain communication between parents, teachers, the school nurse, and school nutrition staff to promote understanding of the student’s complex dietary needs. If necessary, ask the family’s dietitian to come in and train school staff.
- A record of the dietary restrictions prescribed by the student’s physician should be on file with the school nurse and in the school nutrition office. The nurse should notify school nutrition if and when any changes are made to the recommendations.
- Special foods or formulas that meet the student’s nutritional needs may be prepared by the school nutrition staff. No substitutions or alterations to the portion sizes should be allowed. These special meals should be provided at no extra charge to the student.
- If a mistake is made and a child with an inborn error of metabolism is served the wrong food or wrong portion size, the nurse and other school personnel should be notified immediately. Dietary adjustments or medical care may be needed.
- Medical information disclosed to school nutrition staff should remain confidential. It may not be discussed with other children, school personnel, or anyone else who does not have a need to know.

**References**