Special Diets and Inborn Errors of Metabolism

What are special diets and inborn errors of metabolism?

- Inborn errors of metabolism are rare genetic disorders in which the body lacks the protein (enzyme) to turn certain foods into energy normally. Foods that are not broken down to produce energy can build up in a child's system and, in turn, can cause illness, low blood glucose level, intellectual disability, and death.
- The most common inborn error of metabolism is phenylketonuria. However, with the growing capabilities of laboratories to enable DNA testing and a wider spectrum of newborn screening, more metabolic conditions are being identified.

How common are they?

Some of the more common cases of inborn errors of metabolism include

- Phenylketonuria, 1 in 19,000 live births
- Adrenoleukodystrophy (ALD), 1 in 18,000 live births
- Medium-chain acyl–coenzyme A dehydrogenase deficiency, 1 in 23,000 live births
- Homocystinuria, 1 in 39,000 live births
- Galactosemia, 1 in 55,000 live births
- Biotinidase deficiency, 1 in 95,000 live births

What are some common characteristics of children who have special diets or inborn errors of metabolism or of special diets or inborn errors of metabolism as children present with them?

- Most of these children look normal at birth. However, within the first few days after birth or during early childhood, they may look jaundiced and they have either trouble feeding and persistent vomiting or loss of muscle and brain functions.
- They may appear irritable or lethargic.
- Some of these children have a peculiar odor that may range from smelling like maple syrup (maple syrup urine disease) to sweaty socks (isovaleric acidemia).
- Many of these conditions are tested through the newborn screening program and can be diagnosed shortly after birth.
- Once the condition is diagnosed, the baby must be treated by being placed on a very strict diet. This diet prevents the accumulation of any substance that might build up in the child's body and cause harm.
- Often, special infant formulas are used beyond infancy, into childhood. For some children with metabolic disorders, keeping the offending food out of their diets may enable them to grow and develop normally. For others, such as those with ALD, a cure requires a stem cell transplant.

Who might be on the treatment team?

- Children with inborn errors of metabolism require close supervision by a pediatrician/primary care provider in the medical home, a registered dietitian, a geneticist, and a specialist whose work is primarily focused on treatment of people with a specific metabolic disorder. The Care Plan essentially involves dietary measures and should be directed by these professionals with guidance from the child's parents/guardians.
- The Care Plan should outline emergency measures for when these children begin to appear ill.

What adaptations may be needed?

Dietary Considerations

- Meet with parents/guardians before the child's arrival to school or the child care center to discuss dietary measures in detail.
- Meet periodically with parents/guardians, especially at transition times (eg, infancy to toddlerhood, preschool age to school age) to discuss new dietary plans.
- Ask parents/guardians to suggest or provide acceptable treats for their child for class parties and birthdays.
- As children notice differences in what this child can eat, use it as a teachable moment, to discuss nutrition and foods that are good for growing bodies.

Physical Environment and Other Considerations

Many of the children with metabolic disorders require care in distant clinics that specialize in ongoing treatment of their conditions. This requirement results in absence from child care and school, family stress over the cost of travel, missed work days, and the possibility of bad news from the ongoing assessments done to manage the care of these children. Opportunities to keep up with their same-age peers while undergoing their necessary medical care can help.

Special Diets and Inborn Errors of Metabolism (*continued*)

What should be considered an emergency?

- Inform parents/guardians immediately if the child
  - Has repetitive episodes of vomiting
  - Acts unusually irritable or lethargic
- Parents/guardians, together with the treatment team, should provide the indications for calling emergency medical services (911).
- Children with some inborn errors of metabolism may have much more devastating consequences than others when they are ill, and an emergency plan is necessary.

What types of training or policies are advised?

- Dietary adjustments
- Activity restrictions
- Other accommodations

What are some resources?

National Organization for Rare Disorders: https://rarediseases.org, 1-800-999-NORD (1-800-999-6673)

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