MANAGEMENT OF DIARRHEAL ILLNESSES IN PATIENTS WITH BARTH SYNDROME

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Diarrhea Fact Sheet

Although individuals with Barth syndrome are known to have an increased risk for bacterial infections, viral infections associated with diarrhea also present special management problems. To a large degree, it is the reduced muscle mass characteristic of Barth syndrome that dictates special considerations for treating the Barth child (or adult) with diarrhea. Failure to recognize the special medical needs of a child with reduced muscle mass can be life-threatening in diarrheal illness, when the child's reduced muscle mass limits the ability to compensate for the large fluid and electrolyte losses often caused by diarrhea. Having reduced muscle mass also limits the child's nutritional reserve, which is especially important for any protracted illness during which oral nutrition is limited. The hypocholesterolemia that is characteristic of Barth syndrome also may have a role in both the origin and prolongation of diarrhea. Outlined below are several principles to be observed in the management and care of a diarrheal illness in a child or adult with Barth syndrome.

ELECTROLYTES

Muscle not only provides the largest reservoir in the body for potassium, but also serves as the principal regulator of the blood potassium level. Therefore, any individual who has substantially reduced muscle mass will experience more rapid potassium depletion during diarrhea and will require more frequent testing of the serum potassium level. However, even when the serum potassium level is low, patients with reduced muscle mass may become hyperkalemic very rapidly when given intravenous fluids containing potassium. Thus, replacing an apparent potassium deficit with intravenous fluids requires frequent monitoring of serum electrolytes, often three times a day, to avoid hypo-or hyperkalemia. To some degree there can be a similar problem in maintaining a normal serum phosphate level, but this only infrequently becomes clinically important during the first few days of hospitalization unless the serum phosphorus level is depressed by the use of large amounts of intravenous glucose.

NUTRITION

The reduced muscle mass of children with Barth syndrome also limits their ability to handle fasting stress. This is because during fasting or periods of reduced protein nutrition, muscle becomes the principal source of amino acids supplied to the rest of the body to maintain normal rates of protein synthesis in other, more important tissues. Most children and adults with normal muscle mass can tolerate many days of absent or minimal protein intake without significantly impairing protein synthesis needed to fight infections or handle the myriad metabolic consequences of stress. However, in children with Barth syndrome, plasma amino acids drop to levels that seriously impair critical systemic protein synthesis after only one or two days of fasting or inadequate protein intake. As a result, one should begin parenteral amino acid nutrition after 24 hours of hospitalization when the illness is likely to prevent a return to normal nutrition over the next 12 to 24 hours. This can be done as a full parenteral nutrition, which is preferable, or as intravenous supplemental amino acids at the rate of 1 g/kg/d. This increased attention to nutrition is particularly important in Barth syndrome children, who are at an increased risk of infection and for whom, therefore, the maintenance of natural physical barriers to infection, such as the intestinal mucosa, is vitally important.

HYPOCHOLESTEROLEMIA

Hypocholesterolemia is a common characteristic of Barth syndrome, but its cause is not yet known. Because a number of children with Barth syndrome seem to have diarrhea frequently, it is possible that the hypocholesterolemia is caused by increased intestinal losses of bile acids, which are known to cause diarrhea when malabsorbed by the small intestine. However, also possible is that a primary down-regulation of cholesterol synthesis leads to decreased hepatic bile acid synthesis and increased diarrhea due to fat malabsorption. Although Barth children usually do not have classic signs of fat malabsorption, an occasional Barth patient has been found to have this problem. If the cause of a secretory diarrhea is bile acid malabsorption, then relatively small doses of cholestyramine can be extremely effective in controlling the diarrhea. Transient bile acid malabsorption actually is a very common cause in anyone of watery diarrhea associated with viral illnesses, and cholestyramine unfortunately is much underutilized for treatment of watery diarrhea. Although cholestyramine can cause problems such as constipation or formation of intestinal bezoars, such problems rarely develop when cholestyramine is used for short periods of time. If, however, diarrhea in a child with Barth syndrome is caused by fat malabsorption rather than bile acid malabsorption, then that should be easily identified by stool analysis and treated. Also important to recognize is that diarrhea in Barth syndrome may reflect a diminished ability to provide enterocytes with sufficient energy for fluid and electrolyte transport. For this reason also, physicians caring for children with Barth syndrome must pay very close attention to nutrition during acute illnesses, as noted above.

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Terms of Use

Amino acids: any one of a class of 20 organic compounds occurring naturally and are combined to form proteins
Bezoars: a mass formed in the stomach by compaction of repeatedly ingested material that does not pass into the intestine
Cholestyramine: resin used to treat hypercholesteremia. It can bind many acidic drugs in the gastrointestinal tract and prevent their absorption
Electrolytes: various ions, such as sodium or chloride that are required by cells to regulate the electric charge and flow of water molecules across the cell membrane
Enterocytes: the predominant cells in the small intestinal membrane. They are tall columnar cells and responsible for the final digestion and absorption of nutrients, electrolytes and water.
Hepatic: relates to the liver
Hyperkalemic: an elevated blood level of the electrolyte potassium. Extreme degrees of hyperkalemia are considered a medical emergency due to the risk of potentially fatal arrhythmias.
Hypocholesterolemia: the presence of abnormally low levels of cholesterol in the blood
Hypokalemic: the presence of an abnormally small concentration of potassium ion in the circulating blood
Malabsorption: imperfect, inadequate, or otherwise disordered gastrointestinal absorption
Parenteral: taken into the body or administered in a manner other than through the digestive tract, as by intravenous or intramuscular injection
Potassium: a metallic element that in combination with other minerals forms salts that are important in body processes. All body cells, especially muscle tissue require a high content of potassium. A proper balance between sodium, calcium and potassium in the blood plasma is necessary for proper cardiac function.