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Introduction
Synergy between the Metabolic Service, caregiver and Home Nutrition Support Team (HNST) is pivotal in providing optimal nutritional management of consumers, with complex metabolic alterations, who require long-term specialized nutrition support.

This case involves a young male with Barth Syndrome (BTHS) a rare, x-linked mitochondrial disorder. Less than 500 cases of BTHS have been reported worldwide. BTHS is caused by a mutation in the tafazzin gene (TAZ) located at Xq28. This defect results in abnormal modeling of cardiolipin, an essential phospholipid component of mitochondrial membranes that is important in energy metabolism.

As a result, mitochondrial structure and membrane recognition sites are altered leading to impaired function of the mitochondria. BTHS presents with a wide array of manifestations including cardiomyopathy, neutropenia, hypotonia and muscle weakness, fatigue and lack of stamina, growth delay, 3-methylglutaryl aciduria, and an abnormal cardiac profile.

There is no specific treatment for BTHS. Cardiac failure and severe infections are common causes of death in affected individuals. Cardiomyopathy is the cardinal manifestation in BTHS and is usually of the non-compactan variety. Medicines help control varying degrees of cardiomyopathy; however, in many older BTHS patients, transplantation has been necessary. Neutropenia renders the individual more susceptible to infection and diarrhetic illnesses. Bacterial infections can often be effectively treated with antibiotics. Granulocyte colony stimulating factor (GCSF) is often used to stimulate white cell production by the bone marrow.

The Metabolic and HNST RDs worked collaboratively over the past several years as the HPN was continued. The Clinical monitoring occurred in the inpatient setting. Various lab assays were obtained in relation to clinical status and in anticipation of the possibility and potential for developing nutrient deficiencies due to overt malnutrition. Although this patient received a conservative formulation to gradually promote weight gain without cardiac stress, M.V.-13 and MTE-SC were provided daily through the home nutrition support regime. Challenges include multiple infections and hospitalizations associated with his diagnosis, the patient was able to achieve a net weight gain of 30 pounds with the assistance of home nutrition support therapy.

Case Summary
At age 11, a male with BTHS was referred by an Inherited Metabolic Disease Center for home enteral nutrition. The HNST Registered Dietitians (RD) and Metabolic RD and mother conferred regarding the patient’s medical and nutritional status. Consistent with BTHS, the patient had experienced growth delay as a child and had undergone an accelerated growth rate during puberty. Despite this pronounced increase in linear growth, the patient was significantly underweight and malnourished. A very conservative HEN prescription was initiated via gastrostomy (GT) with consideration for potential refeeding sequelae. HEN was continued for two years until the feeding tube had to be removed as a result of recurrent infections. The patient was able to manifest improvement in his nutritional status during HEN despite these recurring medical obstacles, including development of superior mesenteric artery (SMA) syndrome and multiple viral illnesses. Once the feeding tube was removed, his feeding issues remained and his nutritional status further deteriorated. At that point, a decision was made to initiate total parenteral nutrition in the hospital requiring insertion of a port. When stable, this was converted to home parental nutrition (HPN).

The preliminary goal of HPN institution was to achieve cautious weight gain of no greater than 1 pound per week, with a target weight of 140 lbs (20% lower than IBW for non-BTHS males). The weight goal for this patient was to maintain a target weight below 150 lbs to prevent cardiac stress. Additional objectives were to: reduce chronic fatigue related to malnutrition; prevent catheter infections and complications; correct micronutrient deficiencies and optimize potential for cardiac transplantation. Simultaneous medical management of chronic pain, nausea and vomiting and stabilization of cardiomyopathy were also essential.

The Imperial use of a specialized amino acid profile containing supplemental Arginine and Cysteine was implemented based on observations of low Arginine levels in BTHS subjects. It is postulated that patient may be more susceptible to infection and diarrhetic illnesses. Bacterial infections can often be effectively treated with antibiotics. Granulocyte colony stimulating factor (GCSF) is often used to stimulate white cell production by the bone marrow and help fight infections.

Throughout the growth continuum there are numerous nutritional concerns that arise. Painful oral ulcers may ulcerate the reduced muscle mass. In addition to exercise intolerance, reduces caloric requirement and places the consumer at risk for overheating. Careful dietary monitoring is the only way to ensure proper caloric and nutritional intake.

IOP • www.barthsyndrome.org
• www.ninds.nih.gov/disorder/barth/barth.htm
• www.kennedykrieger.org/ki_diag.jsp?pid=2170
• www.hopkinsmedicine.org/cns/Barth_Summary.html

Parasitic infections
Parasitic infections include: (1) toxoplasmosis, resulting in both subclinical and overt disease, with severe sequelae which can be fatal; (2) Strongyloides stercoralis, which can cause intestinal obstruction; (3) Clonorchis sinensis, a liver fluke, which can cause chronic inflammation and fibrosis; (4) Fasciola hepatica, a liver fluke, which can cause biliary obstruction; and (5) Trichuris trichiura, a common intestinal helminth.

Conclusion
The collaboration of the Metabolic Service, HNST and caregiver can foster optimal nutrition care planning and monitoring, as well as facilitate use of the latest advances in HEN and HPN therapy. This alliance serves to promote optimal care and better outcome for the home nutrition support patient with specialized complex metabolic conditions.

References