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MITO 101 - Nutrition

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Key Points

- Mitochondrial diseases are disorders of energy metabolism.
- In infancy and toddlerhood, failure to thrive FTT is a common finding.
- In older children and adults decreased muscle mass and short stature are common findings.
- Good general nutrition is an important part of the care of a patient with mitochondrial disease. There are no specific dietary recommendations or restrictions that work for all mitochondrial disorders. In fact, the most successful dietary interventions are often individualized to the patient and their specific disorder.
- Very few mitochondrial disorders respond well to diet therapy, although pyruvate dehydrogenase deficiency (PDH) and Respiratory Chain, Complex I deficiency may respond to the ketogenic diet.
- Having a dietitian familiar with mitochondrial disorders can be very helpful to patients and their families over time since impaired nutritional status can make mitochondrial dysfunction worse, and conversely progression of mitochondrial diseases can worsen the nutritional status over time.
- It is always important for the primary care provider to assess for, and treat, common infections that might be contributing to the patient's nutritional problems.

Clinical Investigations

- serum quantitative amino acids
- ammonia level
- lactate
- liver function tests
- BUN, creatinine & electrolytes
- complete blood count (CBC)
- total protein & albumin
- prealbumin,
- iron, iron saturation, total iron binding capacity (TIBC)
- folate and B12 levels
- comprehensive fatty acid levels
- thyroid function tests
- growth hormone studies
- adrenalcorticotrophic hormone (ACTH) & cortisol

- serum glucose
- carnitine levels
- urine amino acids
- indirect calorimetry testing

Clinical Features impacting Nutritional status of mitochondrial patients

- Often concurrent gastro-esophageal reflux (GERD), recurrent/cyclical vomiting, gastroparesis, chronic constipation, diarrhea or pseudo-obstruction.
- Many mitochondrial disorders are also associated with hypotonia, developmental delay or other cognitive impairments, and seizure disorders which can have a negative impact on normal food and fluid intake.
- Patients with mitochondrial disorders can develop hematologic disorders and may develop transfusion dependent anemia. In general, bone marrow is a high energy demanding tissue and the development of bone marrow failure signifies progression of mitochondrial disease.
- Although iron is considered an essential nutrient it can act as a free radical and cause ongoing mitochondrial damage. Therefore, iron is supplemented only for documented iron-deficiency anemia in patients with mitochondrial diseases. In mitochondrial diseases, anemia is usually not amenable to dietary interventions. Additionally, some patients with mitochondrial disease do not absorb oral iron well and respond best to IV Iron or Vitamin-iron combinations.
- Thiamine or Pyridoxine has been used to treat Sideroblastic or Pearson's anemia. Some patients may respond favorably to Folate and/or B12 supplementation.
- Exercise intolerance, muscle pain and cramping during physical activities often discourage patients with mitochondrial disease from participating in regular exercise, which improves endurance, strength, muscle mass and energy levels, enhances appetite and decreases insulin resistance.
- Research suggests that structured resistance and endurance exercise programs help patients with mitochondrial diseases and promote their ongoing participation in activities of daily living.

Management of Mitochondrial related Nutrition concerns

- Patients with mitochondrial diseases can develop hypothyroidism, adrenal insufficiency and/or diabetes mellitis type II, which predisposes them to obesity, increases energy demands and further inhibits their ongoing participation in activities of daily living.
- Mitochondrial supplements, seizure medications, GI medications can all impact on micro- and macro-nutrient metabolism or absorption and can induce nausea, vomiting, constipation or diarrhea.
- Close collaboration between the primary care provider and specialists involved in the care of patients with mitochondrial diseases can minimize the negative impact of these factors on the nutritional status of a patient.
- A crucial goal in patients with mitochondrial diseases is to guarantee adequate nutrition. This often requires small, frequent meals, frequent calorie-dense snacks, and pairing complex carbohydrates and proteins which together help maintain steady blood glucose levels.
- The avoidance of fasting especially during times of febrile illness can be very beneficial.
- Gastric and/or jejunostomy tubes are frequently used to supplement calories or provide continuous feeds. Using concentrated infant formulas, protein powders, Duocal, gravies, sauces, whipping or sour cream and oils can be very easy ways to increase calorie intake without forcing large meals on patients with sensitive GI tracts or easy fatigability. Occasionally, patients with mitochondrial disorders do require TPN if they have severe dysmotility or chronic abdominal pain issues.

Mito 101: Nutrition

TABLE 1

SYMPTOM	NUTRITION/FEEDING IMPACT	POTENTIAL TREATMENT
Failure to Thrive	Parental frustration, exhaustion and guilt Decreased alertness & ability to feed Loss of muscle strength & tone Hypoglycemia Dehydration	Increase calorie density of infant formulas Add fats, sauces, gravies, sour cream, peanut butter to foods G-tube placement for supplemental or night-time feeds IV Fluids/hospitalization as necessary Carnitine supplements Periactin or Marinol for appetite stimulation depending on age
Muscle Wasting	Easy fatigue & generalized weakness decrease ability to prepare &/or eat meals.	Small, frequent meals Pre-made or easy meals for low energy days Increase Protein Intake/Protein supplements Protein-Carbohydrate pairing Periactin or Marinol for appetite stimulation depending on age Creatine supplements
Obesity	Decreased mobility Harder to compensate for low muscle tone during meals	Adjust total calories/day Increase lean protein intake Assess for and treat Endocrine abnormalities
Low muscle Tone	Poor head or trunk control Inability to control saliva Difficulty with chewing & swallowing Diminished endurance during meals	Adaptive positioning Adaptive Self-feeding tools Anticholinergics to control saliva G-tube or G-J tube feeds
Recurrent/Cyclical Vomiting	Anorexia Poor intake secondary to fear of precipitating vomiting Poor weight gain or active weight loss	REFER TO CYCLICAL VOMITING SECTION FOR MANAGEMENT
GERD	Poor intake Poor weight gain or active weight loss	REFER TO GERD SECTION FOR MANAGEMENT
Gastroparesis	Recurrent vomiting Abdominal pain Anorexia Pseudo-obstruction	Small, frequent low fat meals, emphasizing liquids Maintain normal blood glucose levels especially in Diabetics Reglan, Eyrthromycin, SSRI's, Buspirone, Ginger, Botox injections

Constipation	Anorexia Nausea & Vomiting Abdominal pain Poor weight gain or active weight loss	REFER TO CONSTIPATION SECTION FOR MANAGEMENT
Pseudo-obstruction	Recurrent vomiting Anorexia/Cachexia	Small, frequent meals Consider TPN Low dose Erythromycin, Reglan
Chronic Diarrhea	Poor intake Poor weight gain or active weight loss	Small, frequent meals Consider Zinc supplementation
Exercise Intolerance	Easy Fatigue Hypoglycemia	Pre & Post Exercise Hydration Carbohydrate-Protein snack prior to physical exertion
Endocrine Dysfunction	Diabetes Mellitis Adrenal Dysfunction Thyroid Dysfunction	Hormone Replacement as necessary Stress Steriods during illnesses Monitor & treat hypoglycemia
Bone Marrow Failure	Chronic anemia increases fatigue causing decreased meal preparation and poor appetite Poor oral Iron absorption	Small, frequent meals Pre-made or easy meals for low energy days Iron containing Vitamin or IV Iron Avoid oral iron supplements Consider Thiamine, Pyridoxine, B12 &/or Folate
Abdominal Pain	Anorexia Nausea & vomiting Increased difficulties with constipation OR diarrhea	Small, frequent meals Chronic Pain management
Neurologic Dysfunction	Cognitive Dysfunction Frequent seizures Low muscle tone	Adaptive positioning Adaptive Self-feeding tools Anticholinergics to control saliva G-tube or G-J tube feeds Consider Ketogenic Diet
Medication Side-effects	Altered absorption between some foods & some nutrients Creation of specific nutrient deficiencies associated with many Seizure medications. High dose Carnitine and Riboflavin (other B Vitamins) increase nausea & vomiting Carnitine can cause diarrhea	Give some medications in between tube feeding boluses Monitor & supplement potential nutrient deficiencies Adjust supplement dosages and/or supplement type based on individual tolerances

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