Diagnosis of mitochondrial disease can be invasive, expensive, time-consuming, and labor-intensive. Therefore, evaluation is not taken lightly. Doctors experienced in diagnosing and treating these diseases will take either a step-wise approach to diagnosis or, in some centers, the evaluation takes place over a few days. The evaluation includes a combination of clinical observations and laboratory tests.

Under ideal circumstances, the evaluation will produce an answer. However, even after a complete evaluation, the doctor might not be able to confirm a specific diagnosis or put a name to the disorder. In many cases, however, the physician will be able to identify which patients do and don't have metabolic diseases.

Mitochondrial disease is diagnosed by:

- Evaluating the patient's family history
- Performing a complete physical examination
- Performing a neurological examination
- Performing a metabolic examination that includes blood, urine, and optional cerebral spinal fluid tests
- Performing other tests, depending on the patient's specific condition and needs. These tests might include:
  -- Magnetic resonance imaging (MRI) or scan (MRS) if neurological symptoms are present
  -- Retinal exam or electroretinogram if vision symptoms are present
  -- Electrocardiogram (EKG) or echocardiogram if heart disease symptoms are present
  -- Audiogram or BAEP if hearing symptoms are present
  -- Blood test to detect thyroid dysfunction if thyroid problems are present
  -- Blood test to perform genetic DNA testing

More invasive tests, such as a skin or muscle biopsy, might be performed as needed and recommended by your doctor.

PROTOCOL FOR PROCEDURES OR SURGERY REQUIRING SEDATION OR ANESTHESIA
(Date)

Re: (NAME)
D.O.B.

(NAME) is a patient with mitochondrial disease with symptoms that include:
______________________________.

Patients with mitochondrial disease can tolerate surgery and anesthesia safely. However, precautions to reduce further the risk from the procedure include:

1. Elective procedures should be postponed if the patient develops any signs of infectious illness around the time of the procedure date;
2. Minimize the time necessary for fasting. The patient should be encouraged to take some fluids (orally or enterally) just before becoming NPO;
3. An intravenous line should be placed pre-operatively and fluids provided until the patient is eating/drinking well, or able to tolerate fluids through a g-tube if present. If the procedure is delayed, IV fluids can keep the patient stable;
4. IV fluids should contain dextrose and electrolytes; do not administer Ringer's Lactate since patients with mitochondrial disease may have disturbed lactate metabolism;
5. For patients with a history of fasting intolerance and/or documented hypoglycemia, or if there is secondary disturbance in fatty acid oxidation, IV fluids should contain 10% dextrose with electrolytes to run at 1.25x maintenance or higher. The higher glucose solution is necessary to minimize catabolism and flux through an impaired fatty acid oxidation pathway. 10% dextrose is more effective than 5% dextrose in accomplishing this goal;
6. If the patient takes any vitamins as part of his/her mitochondrial management, these can be provided once PO/enteral fluids are tolerated;
7. If the patient has a problem with vomiting post-operatively, s/he should be admitted and continued on intravenous fluids until able to tolerate fluids/food.

If there are any questions regarding the safety of induction agents or other anesthetic medications, please consult the following references:


B. 

Please call with any questions.
PROTOCOL FOR SURGERY REQUIRING SEDATION OR ANESTHESIA WHEN EATING OR THE GUT IS DISRUPTED

(Date)

Re: (NAME)
D.O.B.: 

(NAME) is a patient with mitochondrial disease with symptoms that include:
__________________________.

Patients with mitochondrial disease can tolerate surgery and anesthesia safely. However, procedures that impact the gut (at any level) can potentially destabilize these patients.

Mitochondrial disease is often associated with some degree of abnormal gut motility associated with uncoordinated movement or dysmotility; regions of the gut can be affected to different degrees. This can result in problems that can include any of the following: swallowing incoordination, gagging or choking, gastroesophageal reflux, vomiting, delayed gastric emptying, bloating, abdominal pain, constipation and/or incomplete evacuation. Infectious illnesses, surgical manipulation/disruption of the GI tract, and anesthesia can reduce motility further, usually transiently.

In the post-operative period, oral intake of food and liquid diminishes and these patients are at risk for becoming dehydrated. This can occur for several reasons – pain at the surgical site (e.g., as with tonsillectomy), fatigue from the surgical procedure and/or anesthesia, or worsening gut motility with increased gastro-esophageal reflux and/or delayed gastric emptying. Refusal to eat or drink and poor calorie intake can exacerbate fatigue and result in a prolonged recovery and occasionally readmission.

1. Elective procedures should be postponed if the patient develops any signs of infectious illness around the time of the procedure date;
2. Minimize the time necessary for fasting. The patient should be encouraged to take some fluids (orally or enterally) just before becoming NPO;
3. An intravenous line should be placed pre-operatively and fluids provided until the patient is eating/drinking well, or able to tolerate fluids through a g-tube if present;
4. IV fluids should contain dextrose and electrolytes; do not administer Ringer's Lactate since patients with mitochondrial disease may have disturbed lactate metabolism;
5. For patients with a history of fasting intolerance and/or documented hypoglycemia, or if there is secondary disturbance in fatty acid oxidation, IV fluids should contain 10% dextrose with electrolytes to run at 1.25x maintenance or higher. The higher glucose solution is necessary to minimize catabolism and flux through an impaired fatty acid oxidation pathway. 10% dextrose is more effective than 5% dextrose in accomplishing this goal;
6. IV fluids should be continued until the patient is able to tolerate adequate volumes of fluids/food PO/enterally without vomiting;
7. If this patient demonstrates a refusal to eat or drink after 24 hours post-surgery, consideration should be given to providing 1-2 days of parenteral nutrition which can improve calorie intake, reduce fatigue and related symptoms, and shorten the stay in hospital;
8. If the patient takes any vitamins as part of his/her mitochondrial management, these can be provided once PO/enteral fluids are tolerated;
9. For patients with respiratory insufficiency due to trunk muscle weakness, the use of a cough assist to expand their lung capacity has been found to be beneficial.

If there are any questions regarding the safety of induction agents or other anesthetic medications, please consult the following references:
B. 

Please call with any questions.