

## ***Mitochondrial Disorders at a Glance***

Mitochondrial disorders are complex, chronic genetic disorders that occur when the mitochondria (cell level energy producers) in the cell fail to produce enough energy for cells or organs. Mitochondrial disorders typically affect multiple organs and this makes it difficult to predict the severity and/or the range of symptoms in the individual child. There are many different types of mitochondrial disorders and they can vary in their presentation from day to day, hour to hour.

Mitochondrial disorders are more common than had been previously thought and it is now estimated that 1 in 8,500 people worldwide have a mitochondrial disorder.

Mitochondria are responsible for more than 90% of energy needed by the body to sustain life and support growth. Mitochondria are found in all cells except red blood cells. Some mitochondrial disorders affect a single organ, but many involve multiple organ systems and often affect nervous system and muscles.

*Learn more about physical characteristics of Mitochondrial Disorder:*

### **Characteristic findings of mitochondrial disorders**

- Muscle weakness
- Vision or hearing problems
- Liver or kidney disorder
- Diabetes
- Gastrointestinal problems
- Brain problems such as seizures
- Fatigue which may have sudden onset
  - Individuals need to have a place to rest and recharge
  - Individuals will have good and bad days
- Body's inability to properly regulate temperature and stay hydrated
  - Monitor for temperature extremes

### **Specific Mitochondrial Syndromes**

- Leber hereditary optic neuropathy is a disorder that affects the eye.

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- Mitochondrial hearing loss and deafness
- Kearns-Sayre syndrome (KSS) is a condition that affects many parts of the body, especially the eyes.
- Chronic progressive external ophthalmoplegia (CPEO) is a disorder characterized by slowly progressive paralysis of the muscles around the eyes.
- MELAS (Mitochondrial encephalopathy with lactic acidosis and stroke like episodes) primarily affects the brain, nervous system and muscles.
- MERRF (Myoclonic epilepsy with ragged red fibers) affects the nervous system and skeletal muscle as well as other body systems.
- NARP: Neurogenic weakness with ataxia
- Leigh syndrome is characterized by progressive loss of mental and movement abilities (psychomotor regression) and typically results in death within a couple of years.

### **Non-Specific Mitochondrial Disorders**

- Overlapping features of the above conditions can occur.
- Individuals with multiple medical symptoms have been found to have deficiencies in the function of their mitochondria do to genetic changes.

### **Genetics of mitochondrial disorders**

- There are approximately 1,200 people in the United States who have a mitochondrial disorder.
- It can be caused by changes in genes that are found on chromosomes and inherited from one or both parents.
- It can be caused by changes in genes that are only found in the mitochondria and are passed on from the mother.
  - <https://ghr.nlm.nih.gov/mitochondrial-dna>

### **Other possible findings**

- Brain:
  - Developmental delay
  - Mental retardation
  - Dementia
  - Seizures
  - Neuro-psychiatric disturbances
  - Migraines
  - Atypical cerebral palsy

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- Movement disorders
- Strokes
- Autistic features
- Nerves:
  - Weakness (maybe intermittent)
  - Absent reflexes
  - Fainting
  - Neuropathic pain (pins and needles) and burning
  - Dysautonomia (temperature instability and other dysautonomic problems)
- Muscles:
  - Weakness
  - Muscle pain and spasms
  - Low muscle tone and loss of muscle coordination
  - Exercise intolerance
  - May use a wheelchair
- Developmental delays:
  - Learning delays
  - Autism or autism-like features
- Gastrointestinal problems:
  - Pseudo obstruction (the impairment of the intestines in passing food through the intestines.)
  - Irritable bowel syndrome
  - Cramping
  - Diarrhea/constipation
  - Dysmotility (slowing down of bowels)
  - Unexplained vomiting
  - GI reflux
- Kidneys:
  - Kidney disorder
- Heart:
  - Cardiac conduction defects
  - Cardiomyopathy
- Liver:
  - Hypoglycemia (low blood sugar)
  - Liver failure
- Eyes and ears:
  - Visual loss/blindness

- Ptosis (drooping of the upper eyelid)
- Optic atrophy (deterioration of the optic nerve)
- Hearing loss/deafness
- Strabismus
- Retinitis pigmentosa (breakdown and loss of retinal cells)
- Sensitivity to bright lights
- Endocrine:
  - Diabetes and exocrine pancreatic failure
  - Inability to make digestive enzymes
  - Parathyroid failure (low calcium)
  - Hypothyroidism
  - Short stature
- Systemic:
  - Failure to gain weight
  - Fatigue
- Respiratory problems
- Increased risk of infection
  - It may take longer to recover from cold or other illnesses
- Thyroid or adrenal, autonomic dysfunction

## 1. Medical/Dietary Needs

### What you need to know

#### Medical Needs:

- Challenges to maintain health in people with mitochondrial disorder
  - Fatigue
  - Muscle weakness
  - Dysmotility
  - Dysphagia (inability to swallow)
  - Nausea/Vomiting
  - Ataxia
  - Reflux
  - Temperature control
    - Cold stress

- Exposure to cold can result in severe heat loss and trigger an energy crisis
  - Heat stress
    - Especially in those individuals who can't sweat normally
- Avoid toxins
- Cigarettes
- Drugs
- Alcohol

#### Dietary:

Children with mitochondrial disorder need to have a diet with good nutrition to promote growth, development, and energy production. The diet should provide enough protein, vitamins and mineral, and hydration. It is extremely important to **AVOID fasting**.

- Small frequent meals may be better than a typical 3-meal-a-day routine for some children.

## What you can do

#### General guidelines:

- A yearly check-up and studies as needed should occur in the child's Medical Home.
- Be aware of any changes in behavior or mood that seem unusual and notify the parents.
- It is important to be aware of any academic changes. Contact parents when any differences are noticed.
- Be aware, or ask parents, if the child has a medical alert bracelet.
- School age children with may have multiple doctors and specialist visits to monitor medical conditions.
- Dietary Guidelines
  - 4-6 small meals
  - Include a complex carbohydrate and protein at each meal
  - Maintain healthy weight
  - Avoid fasting
  - Vitamins and mineral may help improve mitochondrial function
- Temperature control
  - Wear weather appropriate clothing
  - Avoid extreme cold and heat
  - Be aware of heat stroke and heat exhaustion
  - Consider air conditioning
- Avoid exposure to illness

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## 2. Education Supports

### What you need to know

There is a wide degree of variability in cognitive and adaptive function in individuals with a mitochondrial disorder. Some individuals may have learning disabilities, attention deficit disorders, autism, and pervasive developmental delays (PDD). It is important to have high, but realistic expectations for each child. Because symptoms can vary on a daily basis for a child, is important to maintain constant and open lines of communication between home and school.

Fatigue is a major issue and it is important to be aware of the child's energy level which will vary from day to day. School age children may seem to work in "spurts" and then become lethargic and find it hard to concentrate. Fatigue may lead to difficulty thinking, remembering, and moving.

Consider therapists and specialists to consult and support classroom teacher.

- Physical therapy
- Occupational therapy
- Speech therapy

### What you can do

- Educational supports:
  - Help with organization.
  - Present information in concrete manner.
  - Repetition will help children remember basic facts.
  - Verbal explanations are often more effective than visual demonstrations, diagrams, and models.
  - Provide information in brief, organized, and specific manner.
  - Simplify information presented on worksheets.
  - Tasks like copying letters and figures may take longer.
  - Geometric designs may be difficult.
  - Written homework may need to be modified.
  - Consider having child work with an occupational therapist.
  - A step-by-step approach is most effective.
  - Pay attention to the student's abilities.
  - Remembering locations of objects in space may be challenging (i.e. number lines may be ineffective).

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- Math fractions, geometric shapes, and formulas may be hard.
- Individuals may need help lining up numbers to do calculations.
- Poor executive function can lead to difficulties in calculations requiring more than one step.
- Word problems may be challenging.
- Speech and language:
  - Ensure the child has a reliable way to communicate.
  - Speech and language therapy and supports that are individualized to the child may be helpful.
- Physical Therapy:
  - Develop gross motor abilities.
  - Improve strength.
  - Assess modifications and adaptive equipment for school and home.
  - Muscle cramping and/or spasms can occur as it takes a lot of energy to write, stand in line, write on a chalkboard, and sit at a desk, etc.
- Occupational Therapy can help with:
  - Fine motor
  - Strength
  - Dexterity
  - Fatigue
- Fatigue and temperature control:
  - May need a modified school schedule or rest time/naps.
  - May need to work at home.
  - Extended school year may help prevent regression.
  - Computers may help decrease small motor fatigue.
  - Additional water and snack breaks are needed to remain hydrated.
  - Need to have frequent monitoring of core body temperature, which tends to be lower than the average child.

### 3. Behavioral and Sensory Support

#### What you need to know

Some individuals will have behavioral issues and fatigue may exaggerate these issues. Individuals may benefit from a functional behavioral assessment to identify causes or triggers

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of behavior problems. Some children may have difficulty regulating emotions and behaviors that can stem from living with a chronic disorder.

Some mitochondrial disorders have an increased risk of behavioral issues associated with them including:

- Autism
- ADHD with or without hyperactivity
- Depression
- Bipolar panic disorders
- Anxiety

## What you can do

It is important to treat children who have mitochondrial condition the same as you would with any other child in the class (high expectations, academic achievement) but provide the supports they need to be successful.

Consider:

- A 504 plan
- Behavioral supports
- Counseling
- Medication

Social:

- Provide a safe area to share emotions
- Teach and support social skills development
- Teach about facial expressions, body language etc.
- Misconceptions of abilities and/or limitations can cause insecurity and anxiety
- Help develop confidence and focus on strengths
- If fatigue is an issue, **see FATIGUE section**.
- Identify and use activities in which a child can be successful.
- Encourage socializing opportunities with other children who have a similar interests.
- Develop a plan for addressing emotional concerns. Resources can include: teachers, therapists, and community service providers, in addition to members of the patient's medical team.

## 4. Physical Activity, Trips, Events

## What you need to know

### Exercise:

Exercise has many benefits to the body including; increasing the ability to move, maintaining balance, flexing the joints, improving moods, decreasing depression and anxiety, decreasing the onset of diabetes and heart disorder, increasing energy production in the body and can increase the number of healthy mitochondrial in the cells.

- Moderation is important - don't under or overdo it.
- Exercise tolerance may change day to day. Teach children to listen to their bodies.
- Maintain a supportive environmental temperature.
- Choose activities in which a child can be successful.
- Balance fatigue and rest.
- Time activities to optimize success.
- Hydration is important.
- Avoid exercise during illness or when in a fasting state.
- Some types of mitochondrial disorder have actual weakness not just decreased endurance, so strength exercise may be important and can improve strength.
  - Should have rest between exercises to allow muscles to recover.
  - Resistance exercise (weights) can be performed.
  - Sprinting may be hard because muscles can't regroup and rest.
  - Some individuals may develop rhabdomyolysis (the breakdown of muscle tissue which leads to muscle fibers contents leaking into the blood) when they overdo it.
    - This may lead to painful cramps and muscle breakdown, which may lead to myoglobin from the muscle passing into the urine.
  - See: <http://www.mitoaction.org/blog/exercise>

### Field Trips:

Any change in routine may produce anxiety, fears, and/or worry:

- Offer anticipatory guidance and preparation to prepare for a change in routine such as a field trip.
- Create a picture story about the upcoming event. The child can rehearse it alone or with others.
- If a child has any sensory, hearing or vision issues, he/she may need preferred seating.
- If child is using a wheelchair, make sure transportation and accessibility needs have been considered.

## What you can do

- The amount of exercise an individual can do may vary from day to day with the individual. It is important to find what type of exercise and intensity works best for the child.
- Things to avoid:
  - Cold may result in severe heat loss and trigger an energy crisis.
    - Child should be dressed for the weather when going outside.
    - Child should avoid over exposure to cold for a long period of time.
  - Heat stress may be worse because of the inability to sweat normally.
    - Heat exhaustion and heat stroke may occur.
    - Avoid direct sunlight.
    - Stay indoors and use air conditioner.
    - Encourage hydration and frequent water breaks.
  - Psychological stress:
    - Stress may result in temporary or sometimes permanent worsening of condition.
    - Support the child emotionally.
  - Avoid:
    - Starvation or fasting
    - Lack of sleep
    - Illness, toxins
    - Alcohol
    - Cigarette smoke
    - MSG
- The MitoAction website has an exercise guide to follow ([www.mitoaction.org](http://www.mitoaction.org)).
- For those that live in New England (USA) and qualify, Northeast Passage offers Therapeutic Recreation and Adaptive Sports programming ([www.nepassage.org](http://www.nepassage.org)).

#### Field Trips:

- Offer anticipatory guidance and preparation for any changes in routine.
- Offer supports as needed for vision and hearing issues.
- If there will be lots of walking, the child may need the option of alternative methods of transportation (wheelchair, etc.).

## 5. Absences and Fatigue

### What you can do

Absences:

- Children with mitochondrial disorders may be absent due to surgery, illness and/or medical appointments.

Fatigue:

- Individuals need to learn to conserve energy and accept help from others.

## What you need to know

- Help to make transitions in and out of school as seamless as possible
- Allow extra time for assignments and/or provide make-up work as needed
- Make accommodations for resting or taking break
- Have peers share class notes.
- Monitor work so that it is challenging, but there are attainable and realistic goals.
- Plan for absences and consider tutoring.
- Communication with parents is important to meet these challenges.

Fatigue:

- May be tired and require rest opportunities or breaks.
- Eliminate unnecessary effort.
- Plan ahead.
- Prioritize activities.
- See more at: <http://www.mitoaction.org/blog/dealing-with-fatigue#sthash.Mkk2MH5N.dpuf>

## 6. Emergency Planning

### What you need to know

- Be aware of stress and help child avoid it.
- Have a plan if the child has difficulties due to:
  - Cold stress
  - Heat stress
  - Fasting
  - Fatigue
  - Illness
  - Seizures

## What you can do

- Develop an emergency plan if necessary, depending on the needs of individual children.

## 7. Resources

### Mito Action

Mito Action is a site whose mission to improve quality of life for all who are affected by mitochondrial disorders through support, education, and advocacy initiatives.

- <http://www.mitoaction.org/overview>
- <http://www.mitoaction.org/education-overview>
- <http://www.mitoaction.org/elementary-school>
- <http://www.mitoaction.org/tools-for-special-education>
- <http://www.mitoaction.org/guide/behavioral-issues>
- <http://www.mitoaction.org/blog/psychiatric-disorders-mitochondrial-disease>
- <http://www.mitoaction.org/blog/nutrition-mitochondrial-disease-patients>
- <http://www.mda.org/disease/mitochondrial-myopathies/medical-management>  
<http://www.mitoaction.org/blog/exercise>
- <http://www.mitoaction.org/guide/fatigue-and-exercise-intolerance>

### The United Mitochondrial Disease Foundation

This foundation offers support to people who have mitochondrial disorders.

Teacher/Education Resources

<http://www.umdf.org/site/pp.aspx?c=8qKOJ0MvF7LUG&b=7934659%20>

Treatments and Therapies:

<http://www.umdf.org/site/pp.aspx?c=8qKOJ0MvF7LUG&b=7934635>

### The Foundation for Mitochondrial Medicine

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This foundation supports the development of the most promising mitochondrial disease research and treatments of many forms of mitochondrial disease.

<http://mitochondrialdiseases.org/mitochondrial-disease/>

### **Moving on with Mito: A guide for teens and young adults living with mitochondrial disease**

*"We created this guide for emerging adults living with mitochondrial disorders to help you succeed with transitioning to adult health care and the next phase of life...This guide includes: a simple explanation of Mito for friends, teachers, and anyone else who doesn't understand; tools to help guide your transition to adult health care; tips for success in college, employment, and independent living; and stories of real people with real experiences living their lives to the fullest..."*

[http://www.negenetics.org/sites/default/files/libraries/transition\\_products\\_pubs/moving-on-with-mito.pdf](http://www.negenetics.org/sites/default/files/libraries/transition_products_pubs/moving-on-with-mito.pdf)

### **Including Samuel**

The film *Including Samuel* may be helpful for people interested in including children who have disabilities/health conditions in typical classrooms.

<http://www.includingsamuel.com/>

### **Classroom Accommodations for Students with Visual Issues**

Classroom Accommodations for Students with Visual Issues is a resource from Boulder Valley Vision Therapy, P.C. (<https://www.bouldervt.com/>), in Boulder, Colorado. It describes a variety of variety of issues and offers strategies.

<https://www.bouldervt.com/wp-content/uploads/sites/478/2015/12/227-Course-Handout-CCIRA-2016.pdf>