



Complex Child E-Magazine

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Introduction to Mitochondrial Disorders and Mito Resources

by Susan Agrawal, editor

This month's edition takes a brief look at Mitochondrial Disorders, a collection of genetic diseases that affect the mitochondria within cells by limiting the amount of energy they can produce. Since mitochondria are responsible for creating energy within the cell, thereby supporting organ function and energy throughout the body, defects in the mitochondria lead to a wide variety of chronic conditions with variable symptoms and severities.

How many of our readers have heard of MNGIE, Leigh Disease, or Complex I Deficiency? These are just a few of the many disorders considered to be Mitochondrial Diseases, many of which are listed below in Table 1.¹ While relatively common (about 1 in 3000-4000 births), and perhaps even more common than thought in milder forms, it is only within the past few years that scientists and doctors have begun to focus more on Mitochondrial Diseases. Awareness of Mitochondrial Disorders remains in its infancy for the public at large, and our hope is that this edition will help to provide more information and awareness about these diseases.

Because Mitochondrial Diseases vary so much from individual to individual, it is impossible to list out the symptoms, method of diagnosis, treatments, or prognosis for Mitochondrial Diseases. Instead, one must take the approach of Keely Schellenberg in her article, "Symptoms of Mito: A Surprisingly Short List," a piece that looks at systems of the body instead of individual symptoms when considering Mito as a diagnosis. Melody Pohla's article, "Diagnosing Mitochondrial Disorder: Our Journey through the Genetic Jungle," tackles the difficult area of searching for the specific genetic mutation for her family's Mitochondrial Disorder, providing detailed information on genetics, inheritance, and diagnosis. Finally, Jennifer Peterson's article, "Mito Plus: One Family's Experience with Mito, Autism, and Eosinophilic Disease," evaluates how these apparently unrelated conditions may in fact be closely related for her child and many others.

This edition also offers two poignant commentaries from families dealing with Mito. Cristy Balcells' "Looking Normal" provides a humorous glimpse into day-to-day life for a Mito family. Melody Pohla's "The Pregnancy Journal of a Mito Mom," takes us through being pregnant with suspected Mito, as well as the early days of her three children, two diagnosed with Mito and one likely to be diagnosed in the future.

Table 1: Mitochondrial Diseases

Alpers Disease	LHON
Barth syndrome	LIC (Lethal Infantile Cardiomyopathy)
Beta-oxidation Defects	Luft Disease
Carnitine-Acyl-Carnitine Deficiency	MAD
Carnitine Deficiency	MCAD
Co-Enzyme Q10 Deficiency	MELAS
Complex I Deficiency	MERRF
Complex II Deficiency	Mitochondrial Cytopathy
Complex III Deficiency	Mitochondrial DNA Depletion
Complex IV Deficiency	Mitochondrial Encephalopathy
Complex V Deficiency	Mitochondrial Myopathy
COX Deficiency	MNGIE
CPEO	NARP
CPT I Deficiency	Pearson Syndrome
CPT II Deficiency	Pyruvate Carboxylase Deficiency
Glutaric Aciduria Type II	Pyruvate Dehydrogenase Deficiency
KSS	Respiratory Chain
Lactic Acidosis	SCAD
LCAD	SCHAD
LCHAD	VLCAD
Leigh Disease or Syndrome	

Finally, our Specialty Articles and Features sections contain several articles that are helpful to families dealing with Mito as well as many other conditions. An article on Fluid Requirements comes just in time for the summer heat. Two other articles introducing Palliative Care and discussing the Crisis in Pain Control for Children who are Complex, Non-Verbal, or Cognitively Impaired, provide vital information for families confronting difficult issues resulting from life-threatening diseases and chronic pain.

We hope you enjoy this edition!

Mito Resources

- ❑ MitoAction [<http://www.mitoaction.org/>]: MitoAction is a group providing support, education, and advocacy for families affected by Mitochondrial Disorders. Of particular interest is the MitoAction Blog [<http://www.blog.mitoaction.org/>], which contains both medical and nonmedical articles similar to those found here at Complex Child, but focused exclusively on Mito.
- ❑ United Mitochondrial Disease Foundation [<http://www.umdf.org/>]: UMDF provides medical information, education, and research on Mitochondrial Diseases.
- ❑ Muscular Dystrophy Association [<http://www.mda.org/>]: Mitochondrial Diseases fall under the umbrella of conditions served by MDA. This organization provides a wide variety of supports, including clinics, summer camps, and funding research.
- ❑ Mitochondria Research Society [<http://www.mitoresearch.org/>]: Primarily intended for scientists and physicians, this group focuses on research to find a cure for Mitochondrial Diseases.
- ❑ Parent-2-Parent Mito Support Group [<http://parent-2-parent.com/forum/forumdisplay.php?f=121>]: An online community for Mito issues.

¹ List taken from UMDF at <http://www.umdf.org/site/c.dnJEKLNqFoG/b.3042173>