Mitochondrial Syndromes

Mitochondrial disease symptoms may include impaired hearing and vision, ataxia (challenges with balance, coordination and speech), seizures, learning disabilities, heart defects, diabetes, and poor growth.

Symptoms affect everyone differently and can vary from mild to life-threatening. Younger people tend to have more debilitating conditions. Children with mitochondrial disease may have difficulty developing certain skills such as sitting, crawling, walking, speaking and learning.

Because most people with a mitochondrial myopathy experience symptoms that affect multiple systems at the same time, common symptoms are grouped together and referred to as syndromes. Some of these symptoms are outlined below:

Syndrome	Symptoms
Barth Syndrome *Infancy or early childhood (sometimes adulthood)*	• Enlarged heart • Increased rate of infections • Delays in growth before puberty • Low muscle tone (hypotonia) • Muscle weakness • Specific facial appearance (round face, full cheeks, pointed chin, large ears, deep-set eyes) • Certain laboratory findings, for example: high lactate, low white blood cell count, low cholesterol, increased 3-methylglutaconic acid and 2-ethyl hydracrylic acid in urine or blood, increased monolyso-cardiolipin: cardiolipin ratio • Mainly impacts males
CPEO Chronic Progressive External Ophthalmoplegia *Adolescence or early adulthood*	• Weakness of the eye muscles leading to decreased ability to move the eyes • Ptosis (weakness of the eyelid muscle leading to drooping of the eyelids)
KSS Kearns-Sayre Sydrome *Before age 20*	• Weakness of the eye muscles leading to decreased ability to move the eyes • Ptosis • Abnormal pigment in the back of the eye (pigmentary retinopathy) which may affect vision • Failure to thrive • Abnormal heart rhythm • Ataxia • Certain laboratory findings such as increased protein in cerebrospinal fluid
Leigh Syndrome Note: when inherited through mtDNA, it may be called MILS or Maternally Inherited Leigh Syndrome *Infancy (3 – 24 months) or early childhood*	• Rapid loss of developmental skills, including head control, sitting, standing, or walking following earlier normal development • Decreased level of consciousness • Difficulty breathing • Weakness • Seizures • Low muscle tone (hypotonia) • Ataxia • Abnormal movements • Enlarged heart muscle • Failure to thrive • Certain laboratory findings such as high lactate