PCQD: Primary CoQ10 Deficiency

Mitochondria are parts of a cell that help turn the energy we get from food into energy that the body can use. They are also important in the communication between body parts and creating other materials the body needs. Mitochondrial diseases can cause a variety of symptoms in many parts of the body, particularly those that use a lot of energy like muscles and the brain.

The respiratory chain, also called the electron transport chain (ETC), helps a cell create energy (in the form of ATP) using oxygen. Mitochondrial respiratory chain disorders (MRCDs) are a group of conditions that happen when one of the five major complexes or supporting enzymes of the respiratory chain doesn't work as well as it should.

Coenzyme Q10 is a lipid in the ETC that helps move electrons from complexes 1 and 2 to complex 3 in the ETC. It also receives electrons from other proteins in mitochondria supporting mitochondrial function. Primary CoQ10 deficiency happens when there is a genetic mutation that doesn't allow the body to make enough CoQ10 that's needed by the mitochondria to function normally.

Alternative Names

- Coenzyme Q deficiency
- CoQ deficiency
- Ubiquinone deficiency

Cause and Genetics

There are at least 10 genes known to cause primary CoQ10 deficiency. Most commonly, people with primary CoQ10 deficiency have changes in the genes COQ2, COQ4, COQ6, COQ8A and COQ8B.

Primary CoQ10 deficiency happens when there are changes in both copies of a related gene (autosomal recessive inheritance). Someone who has a change in only one copy of a related gene is called a carrier. If both parents are carriers, there is a 1 in 4 chance with each pregnancy that their child will have primary CoQ10 deficiency.

A related, but separate, condition involving low levels of CoQ10 in the body is called secondary CoQ10 deficiency. People with secondary CoQ10 deficiency aren't born with primary CoQ10 deficiency, but develop low levels of working CoQ10 in their bodies later in life. This can be due to other causes like certain medicines and other conditions like heart failure, kidney disease and diabetes. While this factsheet focuses on primary CoQ10 deficiency, people with secondary CoQ10 deficiency can also have similar symptoms. Please talk with your doctor about the differences between primary and secondary CoQ10 deficiency.

Both males and females can have primary CoQ10 deficiency.

Frequency

Primary CoQ10 deficiency is thought to affect fewer than 1 in 100,000 people.

Symptoms

Primary CoQ10 deficiency causes a range of symptoms. These may differ greatly, even in members of the same family. Always check with your provider if new symptoms appear or you are concerned.

In the most severe cases, signs and symptoms of primary CoQ10 deficiency can be seen shortly after birth, including severe brain dysfunction and muscle weakness, failure of other body systems, life-threatening.

In the most mild cases, symptoms of primary CoQ10 deficiency can begin in someone's 60s. These individuals often have cerebellar ataxia, which causes concerns with coordination and balance.

Symptoms of primary CoQ10 deficiency gradually get worse without treatment.

Symptoms may include:

- Seizures
- Intellectual disability
- Low muscle tone (hypotonia)
- Uncontrolled muscle movements (dystonia)
- Kidney disease (Nephrotic syndrome), which can cause other clinical features including:
  - High cholesterol (hypercholesterolemia)
  - Fluid buildup (ascites)
  - Swelling (edema)
  - Blood concerns including: blood in the urine (hematuria), low red blood counts (anemia), trouble clotting, low white blood counts
  - Weakened immune system and frequent infections
  - Kidney failure (end-stage renal disease)
- Low muscle tone (hypotonia)
- Muscle stiffness and tightness (spasticity)
- An enlarged, weakened heart (hypertrophic cardiomyopathy)
- Uncontrolled, fast eye movements (nystagmus)
- Damage in the nerve connecting the eye to the brain (optic nerve atrophy)
- Damage to the part of the eye that senses light (retinopathy)
- Hearing loss

Diagnosis

Primary CoQ10 deficiency can be diagnosed by:

- A detailed physical examination and medical history, with special attention to neurologic, kidney, eye, and muscle or movement features
- Genetic testing for genes known to cause primary CoQ10 deficiency
- Blood tests looking at the activity of enzymes in the ETC, overall metabolic function and others
- Taking a small piece of muscle to look for changes in the mitochondria and enzyme activity levels(muscle biopsy)

It is important to note that some people with primary CoQ10 deficiency can have normal labs, even people with genetic testing confirming their diagnosis.

Primary CoQ10 deficiency is not included on newborn screening panels.

Treatment and Management

Before beginning any treatment or therapy, please consult with your physician.

There are no established guidelines for managing primary CoQ10 deficiency. There is also evidence that individuals with specific genetic causes of their primary CoQ10 deficiency may respond differently to treatments.

Early diagnosis and intervention are critical in order to limit organ damage. Treatment for primary CoQ10 deficiency typically involves high-dose oral supplementation of CoQ10. Antioxidants may also be used to reduce oxidative stress.

Current treatments for primary CoQ10 deficiency are limited by low-bioavailability, meaning the body cannot absorb CoQ10 well, especially organs like the brain. New therapies are being developed in order to overcome the body's limits of absorbing CoQ10.

Additional treatment and management of primary CoQ10 deficiency is symptomatic and supportive. This may include:

- Feeding support, which may include nasogastric (ng-tube) or gastrostomy tubes (g-tube)
- Early intervention for developmental delays and intellectual disability
- Screening and standard treatment for nephrotic syndrome
- Screening and standard treatment for respiratory insufficiency
- Standard treatment for seizures
- Screening and standard treatment for hearing loss
- Screening and standard treatment for cardiomyopathy
- Screening and standard treatment for vision loss and low vision services
- Standard of care treatment for movement differences
- Physical therapy, occupational therapy, and speech therapy
- Avoidance of mitochondrial toxins like certain drugs, tobacco and alcohol
- Mitochondrial supplements
- Special schooling arrangements

Individuals living with primary CoQ10 deficiency typically work with several healthcare providers regularly based on their symptoms, which may include:

- Neurology
- Nutrition and feeding support team
- Gastroenterology
- Audiology
- Optometry or Ophthalmology
- Cardiology
- Nephrology
- Genetics
- Pulmonology
- Social Work

- Physical therapy
- Occupational therapy
- Speech therapy

It is important that all those living with or caring for someone with primary CoQ10 deficiency have an emergency protocol letter. These letters, which are written and signed by a doctor, share details about prescribed treatment during a crisis and in emergency room settings.

Clinical Trials

For specific details on other clinical trials, visit the MitoAction Clinical Trials page or www.clinicaltrials.gov.

Resources

- Coenzyme q10 deficiency | About the Disease | GARD
- Primary coenzyme Q10 deficiency: MedlinePlus Genetics

Connecting with others who are impacted by a rare disease allows for important information to be shared about day-to day life, prevents isolation, and gives hope. Please contact MitoAction for peer support opportunities at 888-MITO-411 or email mito411@mitoaction.org. Other resources we recommend are:

- New Patient Kit for Mitochondrial Conditions
- Planning and Preparation
- Monthly Expert Series
- Energy in Action Podcast

Sources

- https://www.nature.com/articles/s43856-025-01000-8
- https://www.ncbi.nlm.nih.gov/books/NBK410087/#coq10-def.Evaluation_Strategies_to_Ident
- https://www.mdpi.com/2076-3921/12/8/1652

MitoAction does not provide medical advice, diagnosis, treatment, or legal advice. It is essential that all those living with or caring for someone with a Mitochondrial or FAOD disease have an emergency protocol letter. These letters, which are written and signed by a doctor, share details about prescribed treatment during crises and in emergency room settings. Always check with your doctor if you or your child has concerns as everyone may present with symptoms differently. Before beginning any treatment or therapy, please consult with your physician.