OUR MOST CURRENT “ASK THE MITO DOC” QUESTIONS

Q. I noticed on the mitochondrial site that you should only use medical grade creatine. I am having a hard time figuring out what kind this means. A lot say creatine monohydrate, there is also creatine powder. Can you please let me know exactly what I should look for?

A. Micronized creatine monohydrate is perfectly fine. A commonly available brand is "Optimum Nutrition" but any brand that states the above is ok to use.

Sumit Parikh, MD

Q. How do we know which type of carnitine deficiency we have? Primary/secondary etc. For example: total, free and estrified carnitine are low in both blood and urine so how would we know the specific type?

A. Generally, patients with primary carnitine deficiency have low plasma carnitine levels with total carnitine values often less than 10 with elevated urine values, due to a renal carnitine transport defect. In addition, primary carnitine deficiency can be verified by finding a mutation in the associated gene, SLC22A5.

Sumit Parikh, MD
Q. Can you give HPV vaccine to a child with Leigh Syndrome?

A. Contraindications to vaccinations include allergies to components of the vaccine, previous bad reactions with similar or previous doses of that vaccine, and any form of illness. In mitochondrial patients, that includes minor illness and sometimes even an afebrile illness is enough for me to delay immunizations.

More information in general on the safety and efficacy of vaccines can be found on the CDC website. [https://www.cdc.gov/vaccines/index.html](https://www.cdc.gov/vaccines/index.html)

Fran Kendall, MD

Q. Is Cushing’s syndrome a Mito related symptom or diagnosis?

A. Although adrenal insufficiency is commonly seen in Mito, Cushing’s is not. However, the pathophysiology of Cushing’s syndrome suggest impairment in mitochondrial function. Nonetheless, as stated, it is not a common disease process found in primary mitochondrial disease.

Fran Kendall, M.D.

Q. My child has a biallelic mutation on the IARS2 gene, leading to a mitochondrial disease. She has also lost 1/2 the hair on her head. I want to understand this and would like to know if there are any treatments.

A. Hair loss is not typically a part of these disorders. If they have not yet seen a dermatologist, they should do so. They should also review diet as nutritional deficiencies can cause hair loss.

Sumit Parikh, MD
Q. I have a patient with a partial mitochondrial defect with mutation in the ADRP6 gene with chronic myopathy and weakness who is planning on traveling to Colorado. Are there any special precautions or prophylactic supplements/medications that she can take to help reduce symptoms while she is in CO?

A. There are no clear precautions as most patients tolerate altitude changes okay. Select patients, if their condition worsens, they may need supportive oxygen. No change in supplement regimen is necessary.

Sumit Parikh, MD

Previous Questions Asked in 2019

Q. Do you have any studies or information about the use of Botox in patients with mitochondrial diseases?

A. I would highly doubt that there’s any such study. Nonetheless, Botox injections must be considered in the context of the overall patient. A neurologist who is doing the injections would best know whether a patient would be at higher risk for complications due to their general medical condition or not.

Fran Kendall, M.D.

Q. My doctor thinks I have mitochondrial disease and we are awaiting testing. I am not diabetic, but I am having odd blood sugar symptoms. If I do not get up in the middle of the night and have a snack, I awake the next morning feeling like my fasting blood sugar is very very low, but according to the glucose meter, it is not and is within normal range. It takes me all day for the symptoms to slowly improve and I do not feel right until shortly after my 3rd meal of the day no matter what I eat. I feel like my body isn't using the food I eat properly and I eat more that I should to compensate, yet I am not overweight. My A1C has dropped over the past couple of years from an average of 5.5 to 5.0. I have heard mito and fasting do not mix. What should me next step be?

A. Before we can relate your symptoms to mitochondrial disease, you need to have confirmation of the diagnosis. Your testing shows that your symptoms are not related to
glucose, but other metabolites can be abnormal in mitochondrial disease and vary with fasting or feeding. Once your diagnostic testing is complete, then planning further evaluation and designing a treatment can be addressed.

Gerard Vockley, MD

**Q. I am supplementing with carnitine for angina. I want my cardiologist to test serum carnitine. How long do I have to stop supplementing with carnitine for the blood test to be valid?**

A. We would hold the carnitine for at least a few weeks if measuring it in blood; 4-8 weeks if measuring it in muscle

Sumit Parikh, MD

**Q. I want information regarding the effects birth control has on people with mitochondrial disease.**

A. I don’t have any specific resources that addresses that issue in particular but I suspect it depends on the type of birth control. Of course, some may have absolutely none like the use of condoms whereas others could potentially impact an individual. Again, it depends on birth control and the patient’s underlying disorder and problem set but many women with mitochondrial disease specifically take birth control to help regulate their cycles and prevent some of the autonomic dysfunction and fatigue they get with their monthly menses. It’s a discussion they should have in the context of their problems with their OB/GYN.

Fran Kendall, M.D.

**Q. Is adenosine monophosphate deaminase 1 deficiency a mitochondrial disease?**

A. It’s complicated. Some people think it’s a disease and some don’t. It’s not really a mito disorder, though it does affect energy metabolism. I assume it was diagnosed by a neurologist. If he is having symptoms, he should probably have a full metabolic evaluation. Here is some information about it.


Gerard Vockley, MD
Questions from 2018

Q. I saw a patient in Travel clinic with Mitochondrial type I an IV deficiency; any concerns for malaria prophylaxis with malarone or traveler's diarrhea meds to include loperamide and azithromycin; any other concerns for this patient traveling to Zambia for 1 week?

A. No outright concerns unless they have not tolerated the medications before

Sumit Parikh, MD

Q. I have MIDD (3243a->g mutation) and my hearing loss is progressing. Can anything be done to slow progression or reverse it? Any research trials I should be aware of? Thank you!

A. Unfortunately, no, there is nothing to prevent the progressive hearing loss. In regards to clinical treatment trials, the enrollment criteria and details of the existing trials are variable. My recommendation would be to look for any possible trials for which you might be eligible at clinicaltrials.gov and to update yourself on the status through postings on the UMDF website.

Fran Kendall, MD

Q. I have ptosis but am having problems focusing on my kindle...almost a loss of vision on the perimeter. I do wear glasses and have mito with neuropathy. Please help.

A. Thank you for your question, and I am sorry to hear that your vision loss is affecting your ability to focus and use your Kindle. One of life’s pleasures is a good book! Many mito disorders can affect the eye in a multitude of ways. I would advise that you find a good Neuro-ophthalmologist to help evaluate you and see what might be treatable, such as use of prisms in eye wear. It sounds like you might have CPEO (chronic progressive external ophthalmoplegia).

Some of the eye issues we encounter include: corneal exposure from facial weakness or ptosis repair, limitations of extraocular motility with diplopia, convergence insufficiency with double vision, optic neuropathy with central visual loss, retinal
degeneration with peripheral visual field loss and night blindness or with central visual loss (maculopathy), and cortical vision loss from stroke-like episodes.

Hopefully we will continue to have new medications in clinical trials and eventually approved for mitochondrial disease to stop progression and possible reverse some symptoms. Speak to your primary mito doctor to see if they recommend any additional treatments or supplements at this time. If you do not have one, the UMDF can help locate someone for you to see.

Amy Goldstein, MD

**Q. Do I risk muscle damage if I continue exercising during rhabdomyolysis and atp problems?**

**A.** If a person is actively having muscle breakdown (rhabdomyolysis) - they should not be exercising at that time and wait until their muscles recover. Exercise levels may need to be adjusted until the muscles have fully healed. This is true for muscle breakdown due to mitochondrial disease or other genetic causes.

Sumit Parikh, MD

**Q. My 10 year old niece suffers from Very long-chain acyl-CoA dehydrogenase deficiency; VLCADD.**

The past six months she has had constant diarrhea and throwing up. She has been taken to the hospital and tested in different ways but there is still no solid understanding as to why this is happening. Next they will probe her stomach from both ends.

Her mother keeps her on a very low fat diet, administers regular doses of MCT oil.

She also gives her coconut oil and she drinks orange juice.

Recently, I had heard that coconut oil used regularly and especially in a low caloric diet can cause problems with the body ingesting carbs and almost acts like a lard in the body.

Can you direct me to any information to better understand this?

Also, am I correct that it’s best to maintain an alkaline system and to limit the intake of acids such as orange juice?

As you know it is such a complex condition but I appreciate any guidance, information, suggestions you have.

**A.** There is not a general tendency for patients with VLCADD to have GI issues including vomiting, so the physicians are correct to look for other causes. You don’t say
how much MCT oil the child is on. In excess, MCT oil can cause diarrhea, but usually not vomiting. The primary component of coconut oil is MCT oil, so patients shouldn’t be on both as the fat intake is too high. There is no need to avoid orange juice. I recommend reviewing diet with an experienced metabolic dietitian.

Jerry Vockley, M.D., Ph.D.

**Q. How safe is Plaquenil for Mito patients?**

A. With the exception of medications such as Metformin, generally the decision to utilize a medication or not is based on the severity of the problem one is attempting to treat and whether or not the medication at hand is the only medication to treat that significant problem. Having said that, have the patient discuss the use of any biological such as Plaquenil with their treating physician to determine if it’s use is definitely indicated in the management of their comorbid disease.

Fran Kendall, M.D.

**Q. I have mitochondrial deficiency which has left me with great a deal of neuropathic and myopathic pain. With the opiate situation it has been recommended that I take buprenorphine for the pain, which is debilitating. I know that Buprenorphine has an adverse effect on liver mitochondria...but is it safe to take if liver enzymes are closely monitored?**

A. The general answer is that buprenorphine and other narcotics are not outright contraindicated in mitochondrial disease or dysfunction in most cases. Unless the patient's physician feels otherwise there is no need for special monitoring of the liver just because of mitochondrial disease.

As an aside - we usually do not recommend routine narcotic use for any of our patients - including for neuropathic or myopathic pain.

Sumit Parikh, MD

**Q. My daughter has started her cycle about 3 months ago and has dysautonomia with her mito...she is having some significant swelling (pitted edema) daily that seems to be even worse around her cycle time. She also has hypotension so lasiks are not an option. Her cardiologist says she is "mimicking heart failure" but her cardiac labs and echo are normal...any advice? She also has lots of discoloration (turning blue) in her arms, hands, legs, and feet. Walking when her swelling is this bad is painful for her...**


A. This is not uncommon in Mito:

Is her albumin normal? Is she a candidate for IV albumin infusion?

Can she see a “lymphedema” expert or center?

Wear compression hose or use SCD (compression devices)?

Amy Goldstein, MD

Q. Hi, I am 34 years old and it looks that me and my son both have mitochondrial disease. Unfortunately we live in Poland and doctors don’t know anything about it. I cannot stop blaming myself because I was perfectly healthy all of my life. Before pregnancy, I didn’t eat properly, changed my job and worked all Day and night and overstressed so horrible. When I got pregnant I felt that something is not right with me. When my son was born he had the same symptoms as me when I got pregnant and later on, because my symptoms hasn’t improves till now. My sons are also worse. And my question is it possible that I destroy my mtDNA because of those horrible stress, lack of nutrition, not sleeping enough etc.? I read that mtDNA is very weak and mutate very often during the life especially if they don’t get it what they need. My whole family is perfectly healthy and I also was till the pregnancy and now we- me and my 4 year son are so exhausted. My son spends most of his time lying on the couch because he is too tired. I was also overstressed during my pregnancy. Could I have destroyed my mtDNA during my life and then passed to my son?

A. The simple answer is - yes they likely did not do anything to cause primary genetic mtDNA disease- but the true answer is not as straightforward. We now know that any and all lifetime exposures - from cigarette smoke/nicotine or obesity or diabetes to more dramatic things like cancer/chemotherapy lead to very relevant DNA changes including mtDNA... and these changes affect children and grandchildren if conception occurs after the exposure.

Sumit Parikh, MD