Oxford University Hospitals
NHS Foundation Trust

Mitochondrial Patient Information Evening Friday 6 June 2025

Q&A Session

If you are male with mito, can you pass it onto your children?

This would depend on which gene that is causing your mitochondrial condition and how it is inherited in your family. For example, if a man has a mitochondrial DNA (mtDNA) condition – meaning the altered gene causing his condition is on the mtDNA – then we can be reassuring for his children, as they would not inherit this altered gene. We all get our mitochondria and therefore our mtDNA from our mother's egg and our father's sperm is not thought to contribute any mtDNA. However, if for example a man's mitochondrial disease is caused by having an altered nuclear gene, that is inherited in a dominant pattern, then each of his children would have a 50:50 (1 in 2) chance for inheriting the altered gene and potentially developing symptoms.

You can ask to your specialist doctor or genetic counsellor about the chances for passing on your mitochondrial condition to your children. If we haven't been able to find the gene that is causing your condition through genetic testing, it may still be possible to give you advice around this based on your family history information.

If, as a mum, all my cells are affected with LHON mtDNA3460 will both my sons carry mito disease?

In Leber's Hereditary Optic Neuropathy (LHON) usually all the mitochondrial DNAs are altered (we call this homoplasmy) and we expect all maternal line relatives (whether male or female) to carry the same altered LHON gene in all their mitochondria as well. We don't fully understand why some family members have the altered gene and remain unaffected, and others in the family can go onto develop symptoms. The main symptom in LHON being relatively sudden onset of vision loss, affecting both eyes. Gender, age and smoking all seem to be potential factors for developing vision loss in LHON.

If there is a greater incidence of Parkinson's is there is an increase in Alzheimer's?

Parkinson's disease as we define it is not increased in incidence in mitochondrial disorders, neither is Alzheimer's disease. However, in mitochondrial disease you can get symptoms that can be similar to that of Parkinson's disease and Alzheimer's disease such as tremor or forgetfulness.

With decrease in kidney function, what is the management?

There are many elements to managing kidney disease and that depends in part to the specific type of kidney disorder. In more general terms, ideally kidney function is monitored and, if there is kidney impairment, to be looked after by a renal team.

What bloods are needed (routine) for regular screening and how often?

For most people we recommend yearly full blood count, liver, kidney, thyroid, bone profile, and HbA1c. This might vary slightly between individuals.

Regarding eyes, what should an option be looking for/considering?

There are a few different problems that can involve the eyes in mitochondrial disorders such as droopy eyelids, difficulty moving the eyes, problems with the retina and problems with the optic nerve. These are all managed differently. For most people, we recommend having yearly eye tests and if there are particular issues then you might need to be under a specialist team such as ophthalmology or optometrists.

If our memory gene is not working but our immunity is stronger, does that mean I should have more immunisations?

On the whole, people with mitochondrial disorders can be more susceptible to infections and to suffer with them more severely. We recommend taking up the yearly flu and covid vaccinations to try to reduce the chance of infection. Some people also have the pneumococcal vaccination. This would be our advice regardless of whether your memory is affected or not.

a) Should an infection be treated early and for a longer time for people with m.3243A>G.
b) Can you suggest any antibiotics other than those on the "list".
c) Dentist has tried 2 antibiotics – reacted badly to metronidazole, allergic to penicillin, Doxycycline has not worked.

a) Infection can be viral or bacterial. Viral infections will not respond to antibiotics and therefore advice is to ensure good hydration, regular analgesia and rest. Bacterial infections should treated promptly with a course of antibiotics- 5 day course or 7 day course. It is always recommended to finish course.
b) There have been recent updated guidelines on medications and therefore important to have most up to date list.
c) an organism may be resistant to a certain antibiotic and therefore may require sputum sample or urine sample depending on focus of infection. Therefore, if infection is not improving on an antibiotic that an individual has been prescribed that further testing for antibiotic resistance/sensitivity may be merited.

Is skin cancer or any other types of cancer associated with mitochondrial disease?

We are not aware of an increased risk of skin cancer (or any other cancers with mitochondrial disease) We would recommend that you should seek appropriate referral if there is a strong family history of cancer or you develop any atypical lumps or new symptoms (such as blood in stool, rapid weight loss without dieting). Specifically in terms of skin cancers if you note- increase in size of mole, pigmentation changing, irregular borders etc.

Given the neurological affect of mitochondrial disease, has there been any potential link made to aphantasia?

Aphantasia is the inability to voluntarily visualise mental images. It can be regarded as a neurological variation that affects how people think, dream, remember and learn. There is no known association with

mitochondrial disease and it is not reported to be any more common in those affected by mitochondrial disease.

Should mitochondrial diseases be treated differently due to the need for sugar for glycolic energy production?

Glycolysis is the breakdown of glucose into pyruvate and energy and is vital for cells that require rapid ATP (energy) production. It is the primary source of energy in low-oxygen conditions. Patients with mitochondrial disease should maintain good levels of hydration and nutrition to avoid placing the cells of their body under increased metabolic stress (which we know can occur during dehydration and/or fasting) which can result in increased lactate levels in the blood. High lactate levels can make us feel unwell. If you are unable to eat or drink like you usually would, you should try and maintain your hydration and energy levels by taking an electrolyte replacement solution e.g. dioralyte, electrolade and seeking medical advice. Mitochondrial patients who have been unable to tolerate their usual diet or fluids for 24 hours should be reviewed by the GP or local hospital team to ensure they are not dehydrated or running low blood sugar levels.

I have been invited to participate in a Precision Study, as my DNA tests have not identified the gene causing my health problems. In order to make an informed decision, it would be helpful to know whether anyone present has undergone such a study, how they found it and if willing reveal results.

The Precision Medicine Diagnostics Study is a UK based research study funded by The Lily Foundation. The study aims to diagnose individuals with suspected mitochondrial disease who haven't yet received a genetic cause for their difficulties via NHS testing. The study also aims to help accelerate the adoption of advance genetic technologies into the NHS. This is a relatively new study and patients from Oxford are only just starting to be invited to take part. There are no results to share as yet. If your medical team think you are eligible for the study, you will be contacted in due course and provided with an information sheet about the study and the opportunity to ask any questions before you give your consent to take part. Patient support groups, such as those run by The Lily Foundation, can often help connect patients who have been involved in research studies to share their experiences of taking part.

Can nothing else but genetic mutations/variants cause mitochondrial malfunction? How is that substantiated scientifically?

Primary mitochondrial disease is a genetic disorder. Secondary mitochondrial dysfunction has many potential causes and not all will be genetic in origin. As scientific advances progress, we are learning more about the mitochondria and the damage that can be caused by acquired causes. We know that the normal ageing process can damage mitochondrial DNA and impair function, along with many other causes of cellular stress which can contribute to mitochondrial dysfunction. However, it is important to understand that these conditions are not primary mitochondrial disorders and will have specific management to address the underlying cause, unlike primary mitochondrial disorders for which there is currently no treatment or cure.

Will the rare mito disorders service look to have more joined up care for patients? A friend with Multiple Sclerosis (MS) has a MS Nurse to organise her care and to be the first point of contact.

There are currently three specialist mitochondrial services in England commissioned by NHS England Highly Specialist Services. Each of these Services do have a Clinical Nurse Specialist (CNS), along with several other members that make up a multidisciplinary team alongside the doctors, but it is not possible for the CNS to be the first point of contact or organise your care locally due to the number of patients seen by the three services. Maintaining a good relationship with your GP and local hospital team is vital and we will copy them into your clinic letters when we review you. They are welcome to contact us directly if they have any questions or queries about your condition, or a change in your clinical state. There are also clinical guidelines that they can access for further advice and guidance.