The Lily Foundation
Guide to Mitochondrial Disease

Helping you understand mito and find the support you need


Tom Briggs
Guide Dogs
Guide dog

Contents

This resource has been developed by The Lily Foundation in response to feedback from the mito community, recognising the need for clear information at point of diagnosis.

Reviewed by experts, it offers general information to help improve understanding of mitochondrial disease. All content was believed to be accurate at time of printing, but details may change without notice and are beyond the control of The Lily Foundation.

This guide is not a substitute for medical advice, and The Lily Foundation cannot accept responsibility if used as such. Every case of mitochondrial disease is unique, so please consult your specialist or NHS clinician about any individual health concerns.

Hello,
If you're reading this, you or someone you love has just been diagnosed with mitochondrial disease. I wish I could sit with you right now, hold your hand and tell you that everything you're feeling is completely valid.

When I was in your shoes, I felt as if the ground had been pulled from under me. A disease I'd never heard of had just changed my family's life forever. Despite an incredibly supportive clinical team looking after us and Lily, there was no clear plan, no one to give us the answers we needed and no way of knowing what tomorrow would bring. It was lonely, confusing and frightening.

You might feel like no one around you really understands - even some of the doctors, family members or friends you thought you could rely on. You might feel grief for the life you imagined or the life you used to have. Please know this: you're not alone.

At The Lily Foundation, we exist to walk beside you through those early days and beyond. We can help you find your footing, we can answer your questions and be there for you when the world feels too heavy. You're now part of a community of over 1500 families who "just get it."

We're here to offer you both emotional support and practical advice. Together we can:

- Help you navigate the uncertainty of those first days
- Support you as you learn to live with mitochondrial disease
- Offer spaces to connect, from virtual coffee mornings to residential weekends
- Provide counselling and emotional support when you need it most
- Help make special memories through our Lily Wish Fund.

From this moment on, you don't have to face this journey alone. We're here to hold your hand, guide you and give you hope.

With love,
Liz Curtis MBE
The Lily Foundation CEO, Founder and Lily's mum

Small but mighty

The role of mitochondria in health and disease

What are mitochondria?

Mitochondria are tiny structures (or organelles) that are found in almost every cell in our body.

Despite their small size, mitochondria play many crucial roles in the human body, but one of their most important jobs is to transform the food we eat into the energy needed to allow our cells to work and thrive.

That's why mitochondria are often referred to as the 'powerhouse' or 'battery pack' of the cell.

When groups of cells work together, they form tissues and organs, which means the energy produced inside each cell ultimately powers the function of entire organs and systems in the body.

Within each cell there may be hundreds or even thousands of mitochondria, with organs that require the most energy to work (such as the brain, muscles and heart) requiring more mitochondria to provide the energy they need to function.

"We adore our Lily family, the one place where we can just be us! We are understood and listened to here."

Family affected by mito


Understanding
ATP
COMPLEX
CELLULAR INTERACTIONS
VITAMINS
400 DISEASES
mtDNA
FOOD
CARBS
FAT
ATP
Prof Bobby McFarland

What is mitochondrial disease?

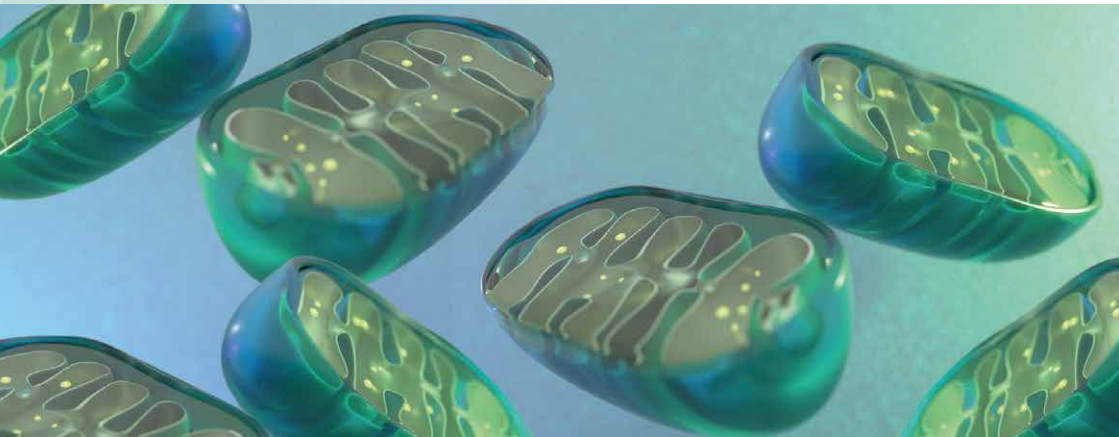
Mitochondrial disease is an umbrella term for a group of genetic conditions that affect the body's ability to transform food into energy.

This failure to produce energy means that the body's cells are unable to function as they should, leading to a wide range of symptoms that may include vision and hearing loss, muscle impairment, seizures and stroke-like episodes.

As the cells fail, the organs that require the most energy to work, including the brain, muscles and heart, stop working as they should or, in severe cases, even shut down. They're often progressive diseases, meaning that symptoms will worsen over time.

There are very few treatments, and currently no cure, for any form of mitochondrial disease, though scientists, research teams and patient organisations including The Lily Foundation are working hard to change that.

Mitochondrial disease is classed as a genetic condition because it's caused by a fault present in a person's DNA at birth, though symptoms may show up at any age.



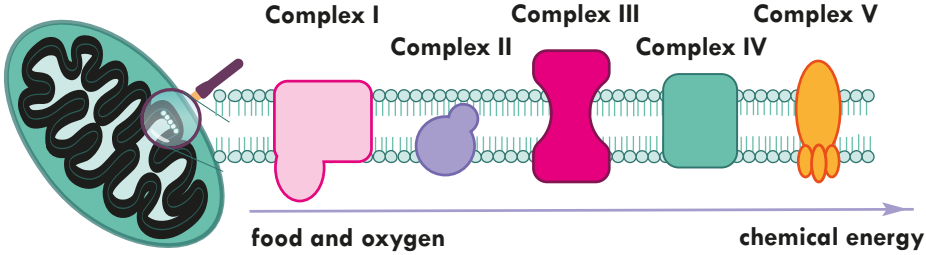
How do genetic faults affect energy production?

You may have heard your doctor refer to the term OXPHOS (short for oxidative phosphorylation) during your clinic appointments. OXPHOS is the name for the process your mitochondria use to transform the food you eat into a form of energy your body can use. This usable form of energy is called Adenosine Triphosphate, or ATP for short.

OXPHOS is made up of five protein complexes, called Complexes I, II, III, IV and V. Complexes I – IV are collectively known as the mitochondrial respiratory chain or electron transport chain, and they work together to pass energy along the chain. Complex V (also called ATP synthase) then uses the energy stored by the respiratory chain to make ATP. Each complex is built from proteins (tiny building blocks in our cells that do important jobs) that work together to pass energy along the chain.

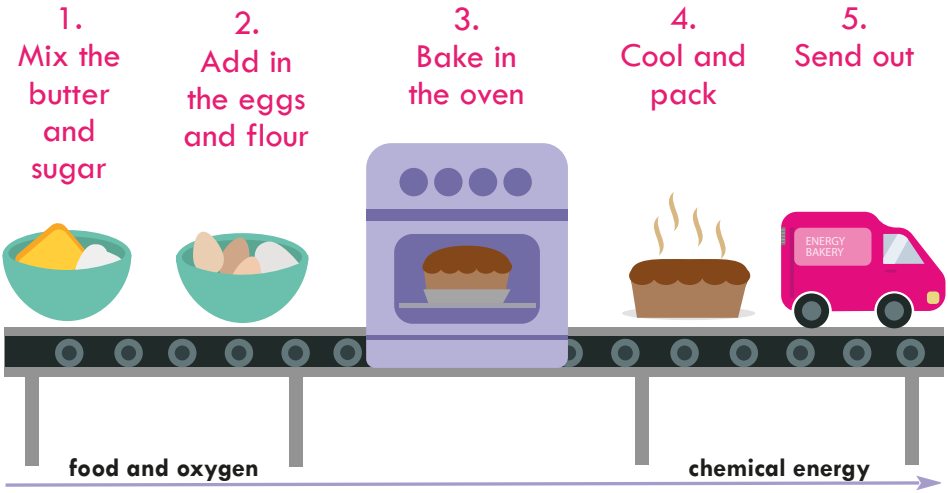
Together, these complexes act like pieces of machinery that turn food into energy. If there are faults in a person's DNA, this can disrupt the way OXPHOS works, making it harder for cells to produce enough energy. This is what leads to mitochondrial disease. The OXPHOS process is pictured below. You can see the complexes marked out and the production of ATP at the end.

Electron Transport Chain



One easy way to imagine how OXPHOS works is to think of it as a cake factory production line. Each stage of the line has an important job to do, just like each complex in the respiratory chain.

If one stage goes wrong, the finished cake will come out wrong, and in the same way, if one complex doesn't work properly, the cell cannot produce energy correctly.



This last stage is just like the final step of the chain, when the mitochondria release ATP, the energy our cells can actually use.

If any step in this process is missed or doesn't work properly (for example, if the eggs are left out or the oven temperature is wrong) the final cake may be smaller than expected or undercooked.

It's the same with OXPHOS. Each complex has its own important job, and if one of them doesn't work properly, energy can't be produced in the right way.

And just as a spoiled cake won't taste as good as a perfectly baked one, faulty energy production means the body can't work as it should, leading to the symptoms of primary mitochondrial disease.

There isn't a simple link between which part of the respiratory chain has gone wrong and how severe a person's disease will be.

This is because the effects depend on many other factors such as how much energy different organs need, how many cells are affected, and how well the mitochondria can still make energy.

In addition, some people with mitochondrial disease can even have more than one complex affected. For example, you may be told that Complex I and Complex IV are both affected, but this doesn't necessarily mean the disease will be more severe.

Not everyone who has mitochondrial disease knows exactly which part of the respiratory chain is affected.

Muscle biopsies can be extremely helpful tools in diagnosing mitochondrial disease, because doctors can tell by looking at a muscle biopsy which of the mitochondrial complexes aren't functioning correctly. However, it won't pinpoint the exact problem.

For this, genetic testing (see page 29) is needed to identify the precise gene responsible for causing disease, and these methods are increasingly being used as the first-line approach to diagnose mitochondrial diseases.


DELL

What goes wrong in mitochondial disease?

Mitochondrial genetics and inheritance patterns

Understanding the genes behind mitochondrial disease

One of the many complicated things about mitochondrial disease is that it can be caused by faults in many different genes, across two different sets of DNA.

Up to now, scientists have found over 350* genes that may be responsible for causing mitochondrial disease, with new ones being found all the time.

It's important to know exactly which genetic fault has caused mitochondrial disease in order to understand the disease better, access clinical trials and eventually enable treatments for mitochondrial disease.

For families considering their options for having children, it's also very important information in order to make reproductive choices.

"Not many people understand what mitochondrial disease is, so it is so nice to be able to chat to people who understand and may be experiencing similar. It makes me feel less alone. The Lily team are lovely and supportive."

Clare, adult with mito

* At time of print - 2025

Nuclear and mitochondrial DNA

You may remember from your biology lessons at school that we all have our own unique genetic 'blueprint' which provides the code that makes us who we are.

This is known as our nuclear DNA, and it contains the vast majority – between 20,000 and 25,000 – of our genes. The genes found in our nuclear DNA provide the instructions that make up all of our unique traits, from the colour of our hair and eyes to how our bodies function.

Nuclear DNA is housed within the cell's nucleus, where it's tightly coiled into structures called chromosomes. You may recognise the double helix shape of DNA, this is where our genes are found, like instructions written along the strand.

Lots of people do not realise that we also have a second, much smaller set of genetic information, and this is stored in the mitochondrial DNA.

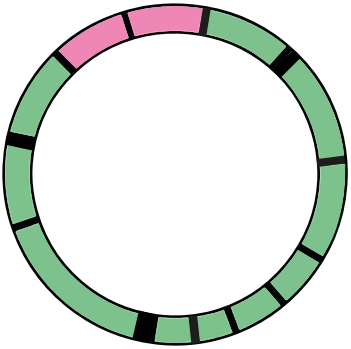
This circular structure, found within the mitochondria, only contains 37 genes and provides us with a tiny amount (around 0.1%) of our genetic information.

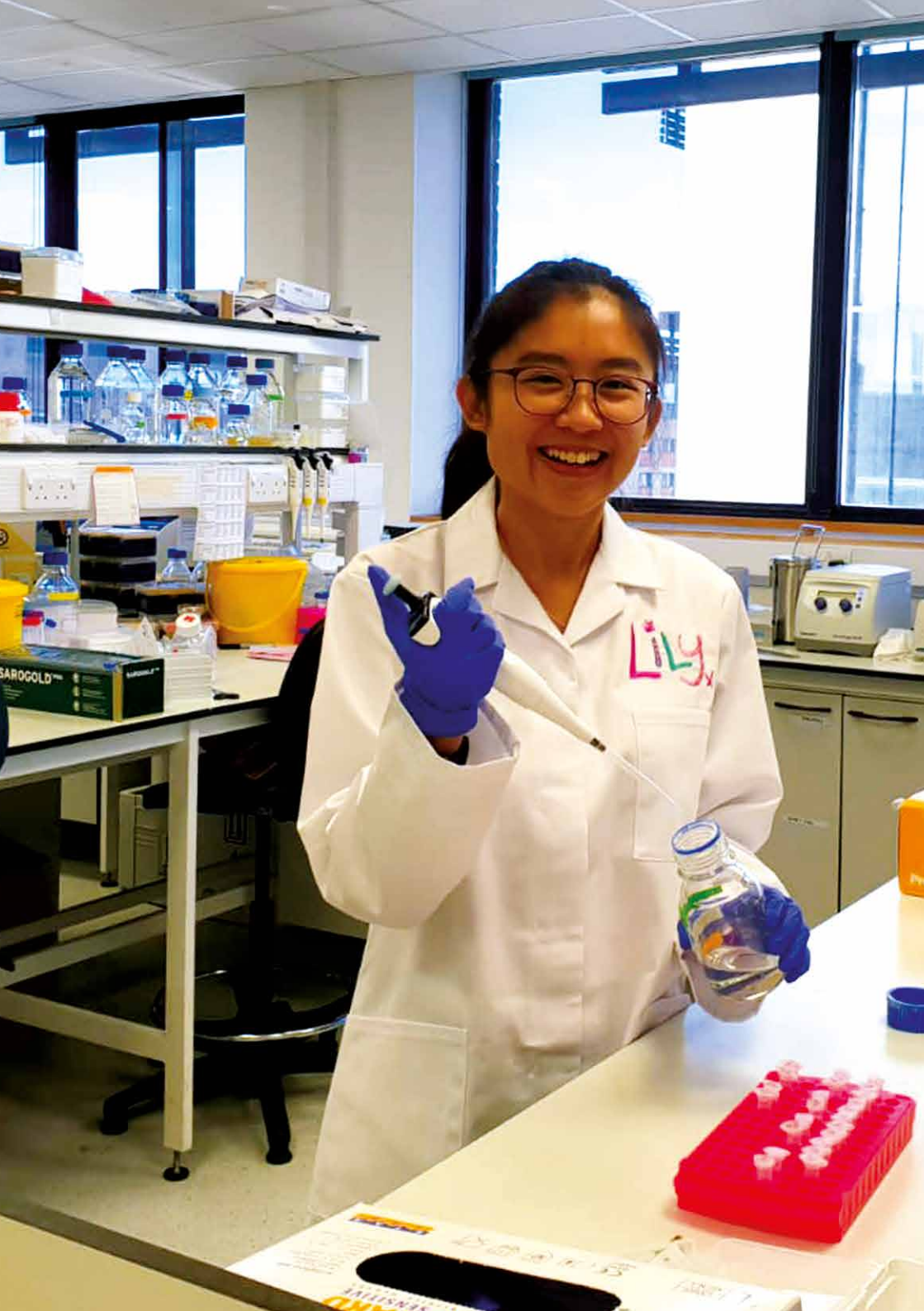
However, this second set of DNA plays a crucial role in energy production.

Another thing that makes mitochondrial DNA different is that it is only passed down from our mothers, in a pattern known as maternal inheritance.

Nuclear DNA

Mitochondrial DNA




Lily x


Lab

Mitochondrial disease inheritance

Mitochondrial disease can be caused by genetic faults in either our nuclear or our mitochondrial DNA and can be inherited in various different ways. This is one of the things that makes diagnosing mitochondrial disease so difficult, because scientists need to look across both our nuclear and our mitochondrial DNA to try and find the root cause of the problem.

It's essential to understand how primary mitochondrial diseases are inherited because this has important implications, not only for the person affected by mitochondrial disease, but also for the wider family and any future generations.

Mitochondrial disease can be inherited in various ways, depending on whether the mutation is found in the nuclear or the mitochondrial DNA.

In **nuclear DNA mutations**, there are two main inheritance patterns: **autosomal recessive and autosomal dominant. Approximately 75%** of **paediatric presentations** of mitochondrial disease are caused by mutations in the nuclear DNA.

In **mitochondrial DNA mutations**, these are passed down the maternal line, known as maternal inheritance. **Approximately 80%** of **adult cases** of mitochondrial disease are caused by mutations in the mitochondrial DNA.

X-linked inheritance is another pattern, where the mutation is located on the X chromosome. Because males have only one X chromosome, they're more likely to be severely affected, while females may have milder symptoms or sometimes act as carriers. X-linked mitochondrial diseases are much rarer than the other inheritance types.

In addition, both **nuclear and mitochondrial DNA mutations can happen sporadically** and may be known as 'de novo' (meaning 'new') mutations.

Overleaf, we'll focus on **autosomal recessive, autosomal dominant and maternal inheritance**, as these are the most common patterns.

Autosomal recessive inheritance

Autosomal recessive inheritance occurs when the fault is in the nuclear DNA. This is where the person affected by mitochondrial disease needs to have two faulty copies of a gene in order for it to cause disease.

We know that nuclear genes are inherited in pairs. If both Mum and Dad have a single faulty copy of a gene that's capable of causing mitochondrial disease, they're known as carriers.

Because they have one healthy copy of the gene, they'll be protected from having symptoms.

When Mum and Dad go on to have children, we know that they'll pass a single copy of each of their genes down to their children. There are a few possible outcomes here:

If both Mum and Dad pass down their healthy copy, the child will also be healthy and it's highly unlikely that they'll go on to develop a mitochondrial disease in the future.

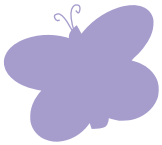
If both Mum and Dad pass down their single faulty copy to their child, that child will inherit two faulty copies and therefore will be affected by mitochondrial disease.

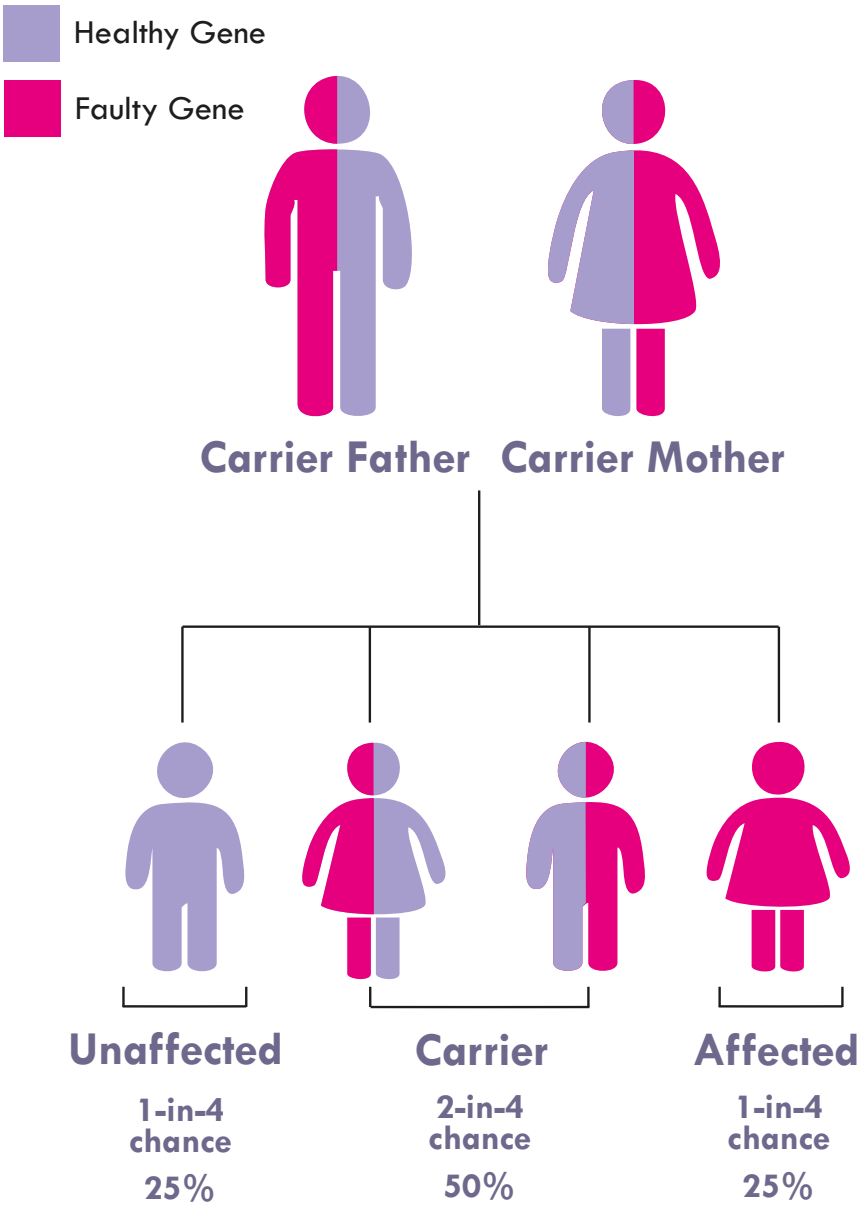
If either Mum or Dad passes down a faulty copy, but the other passes a healthy copy, then that child will be a carrier of disease, but they won't have symptoms themselves.

However, they do have a risk of passing this on in the future when they go on to have children of their own.

In autosomal recessive inheritance, there's a 1-in-4 chance that the child will inherit two faulty copies and go on to develop mitochondrial disease.

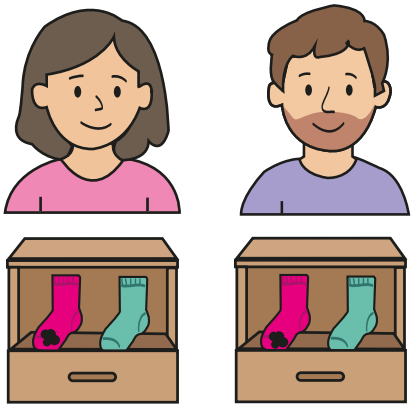
There's a 1-in-4 chance that the child will inherit two healthy copies, and a 2-in-4 chance that the child will be a carrier.




Healthy Gene
Faulty Gene
Carrier Father
Carrier Mother
Unaffected
1-in-4 chance
25%
Carrier
2-in-4 chance
50%
Affected
1-in-4 chance
25%

It can be easier to think about autosomal recessive inheritance by using the 'sock drawer' analogy.

Imagine each parent has a sock drawer with two socks in each, one pink and one green.

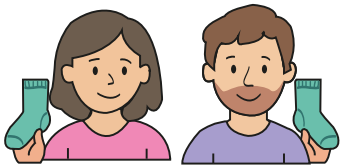


The green sock works fine (to represent the healthy gene). The pink sock has a hole in it (to represent the faulty gene).

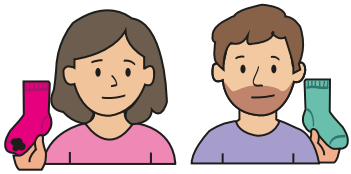
Because both Mum and Dad have one 'healthy' green sock, they are protected from disease.

But when the parents decide to have a child, both parents pass down one of their socks at random.

There is a **1-in-4 chance** that both parents pick out the green sock. That means the child is not affected and is not a carrier.

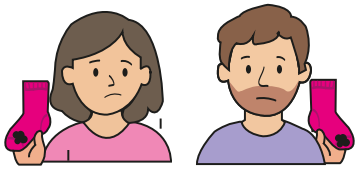


There is a **2-in-4 chance** that one parent picks out the pink sock and one picks out the green one.

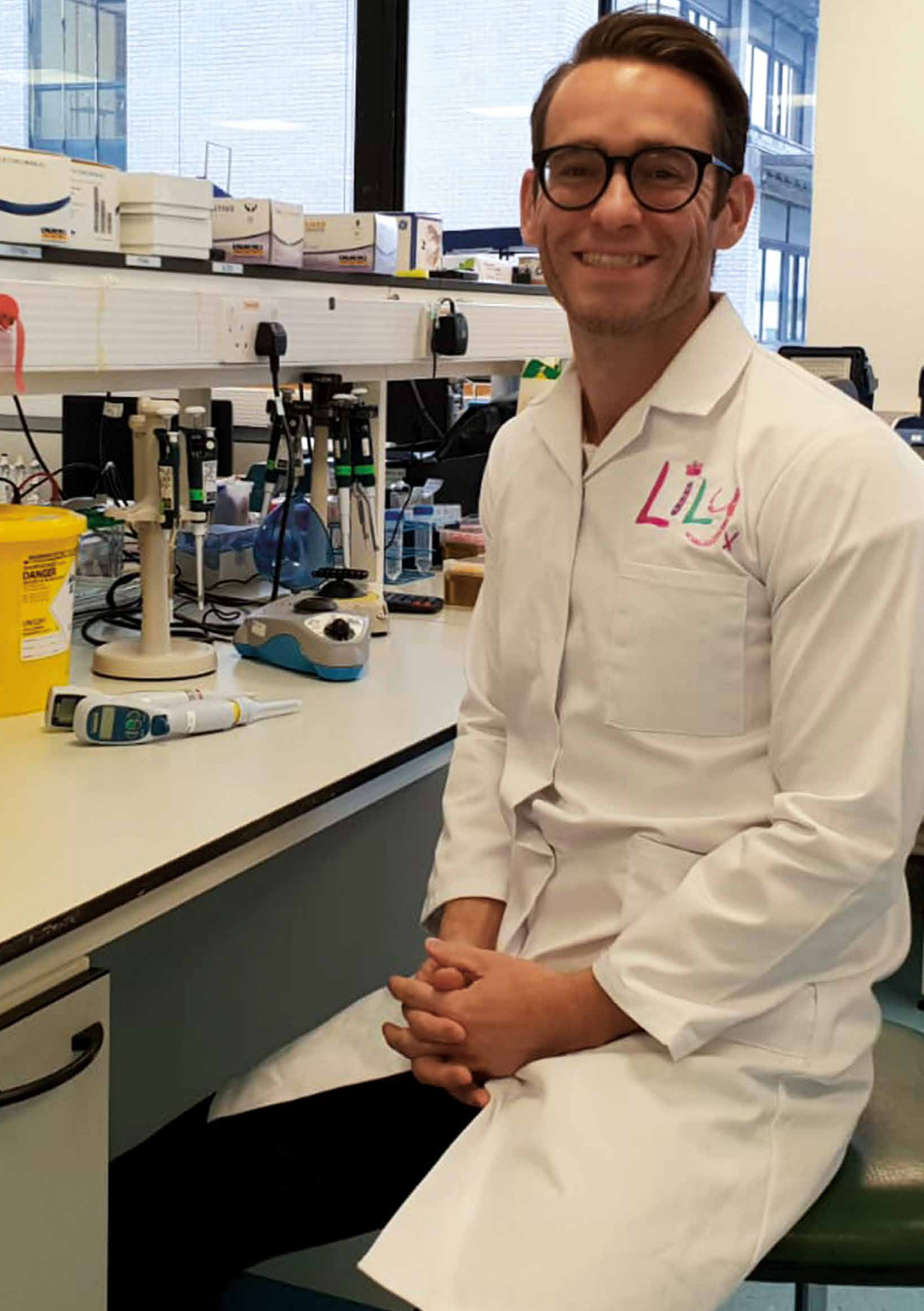


That means that the child is a carrier, so they're not affected but could pass it on.

There is a **1-in-4 chance** that both parents pick out the pink sock.



In this scenario, the child does not have a healthy green sock to protect them, meaning that they will be affected by the condition.


DANGER
Lily x

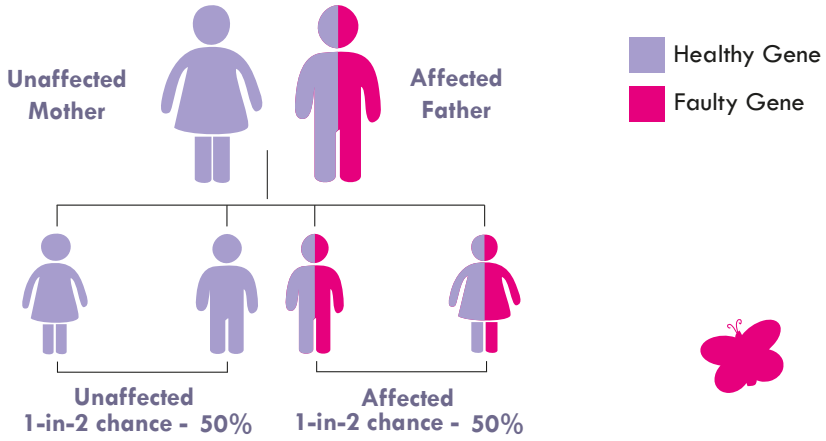
Autosomal dominant inheritance

Autosomal **dominant** inheritance and autosomal **recessive** inheritance may sound the same and both occur when the fault lies in the nuclear DNA. However they represent **completely different** patterns that can lead to genetic disorders.

Autosomal dominant inheritance only needs **one** faulty copy of a gene to cause disease, so the pattern of inheritance is different. (The one faulty gene is dominant - hence the name.)

If either parent has one faulty copy, they could be at risk of having disease themselves and of passing it on to their children.

This time the graphic looks like this:

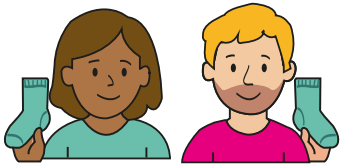


So, going back to our sock analogy, in autosomal dominant inheritance, if there's just one pink sock between Mum and Dad that has a hole in it, that's enough to cause disease.

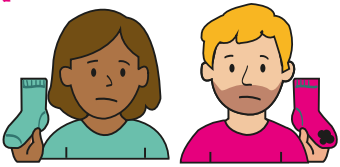
In this scenario, Mum has two green socks, meaning she has two healthy genes and will always pass on a healthy green sock.

However, if Dad has one green sock (a healthy gene) and one holey pink sock (representing the faulty gene), that's enough to cause disease.

When they have a child, there's a **1-in-2 chance** he'll pass on his green sock, in which case the child will have two healthy copies of the gene and won't be affected by the disease.



However, there's also a **1-in-2 chance** that Dad passes down his pink sock.



If this happens, the child will inherit the faulty gene and is at risk of developing disease.

Autosomal dominant presentations of mitochondrial disease are generally thought to be less common than those passed down in an autosomal recessive manner. However, there are some examples of conditions inherited this way, such as Autosomal Dominant Optic Atrophy, a type of mitochondrial disease that mainly affects the eyes.

It's also important to note that the degree to which someone who is affected with an autosomal dominant condition can vary, meaning that people within the same family may be affected by the condition to varying degrees.

It's not possible to assume that if the parent is mildly affected, their children will be mildly affected too.

In terms of inheritance 'risk', for both autosomal recessive and autosomal dominant conditions, the inheritance pattern remains exactly the same for every pregnancy, meaning that if you have a child with mitochondrial disease, you have the same chance that any future children will also be affected.

Maternal inheritance

This means that if a mother has a mutation in her mitochondrial DNA, all her children will inherit it, regardless of whether they're boys or girls.

However, this doesn't necessarily mean that all her children will be affected in the same way.

This is because each of our cells contain many, many copies of mitochondria, and in cases like this, some will carry the mutation while others won't.

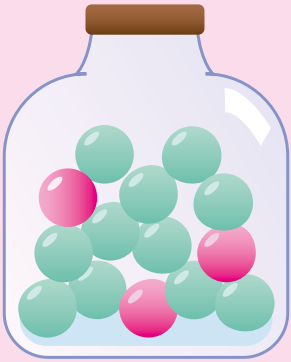
This is known as **heteroplasmy** and helps to explain why mitochondrial disease can affect people very differently, even within the same family.

One child might have no symptoms, whilst another may be severely affected, depending on the proportion of faulty mitochondria in their cells.
To help us understand **heteroplasmy** a little better, imagine every cell in our body as being like a jar of marbles.

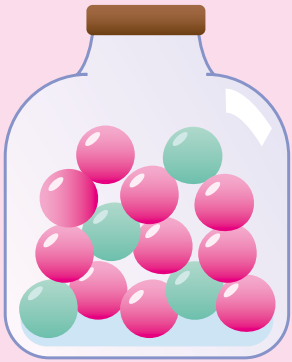
Each marble in the jar represents one copy of mitochondrial DNA inside that cell.

Some of the marbles in the jar are green, whilst some are pink. The green marbles are healthy mitochondria, but the pink ones are the mitochondria that have a mutation that could cause disease.

If the jar has mostly green marbles, the cell can function fairly well.



However, if the jar has mainly pink marbles, the cell will not work properly.



Because different cells can have different amounts of pink and green marbles, some parts of the body might be more affected than others. If someone from the same family has a different combination of pink and green marbles, this helps to explain why members of the same family are affected differently.

Understanding the risks of maternal inheritance in pregnancy

We can also use the marbles in a jar analogy to think about risks during pregnancy. This time, the jar represents all the mitochondria inside one of the mother's eggs.

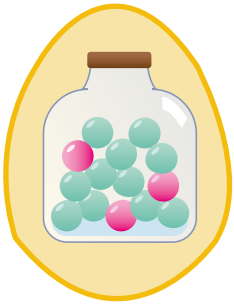
Green marbles are healthy mitochondria. Pink marbles are mitochondria with a mutation.

Each egg can have a different mix of green and pink marbles.

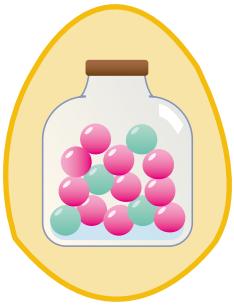
Some eggs might have mostly green marbles (mostly healthy mitochondria), while others might have more pink marbles (more faulty mitochondria).

If the egg has mostly green marbles, the baby is likely to inherit mostly healthy mitochondria and may have fewer or no symptoms.

When an egg with a higher number of pink marbles is fertilised, the baby has a higher chance of inheriting more faulty mitochondria and could be more likely to develop symptoms of mitochondrial disease.



Lower chance



Higher chance

Because every egg can have a different mix, it's very hard to predict exactly what the outcome will be. This is why the risk of having a child affected by mitochondrial disease can vary, even for the same mother.

There are several options for women with mitochondrial mutations who are considering starting a family, so speaking with a specialist about these is crucial.

Sporadic or 'de novo' mutations

Not all mitochondrial diseases are inherited from parents.

There are some mitochondrial diseases that occur spontaneously. These are sometimes referred to as sporadic or *de novo*, which means 'new'.

These types of mitochondrial disease are still genetic in nature, because they've been caused by a change in a person's DNA, but it's the first time the change has appeared in that family.

The types of mitochondrial disease most commonly caused by sporadic mutations are single large-scale deletions of mitochondrial DNA (mtDNA).

Examples include Pearson syndrome, Kearns-Sayre syndrome (KSS), and chronic progressive external ophthalmoplegia (or CPEO).

However, sporadic mutations can happen in both the nuclear and the mitochondrial DNA.

Getting a diagnosis of mitochondrial disease

As we've already explained, finding the exact genetic cause that confirms a person's mitochondrial disease can be extremely challenging, and some people wait many years for scientists to identify theirs.

The process has been compared to looking for a needle in a haystack or a single spelling mistake in an entire dictionary.

Identifying the precise genetic change requires multiple specialist techniques, and you'll find the key steps explained in this chapter.

"You feel so helpless when you get the diagnosis and you're told there's no effective treatments or cure… But there's ongoing research, so you just want to put money in that pot. I'm motivated by the fundraising now and that keeps me mentally and physically fit. It's also good preparation for what's ahead of us.

This is our path now. It gives us purpose. It helps us channel pain into something positive and feel like we're contributing to research that could change lives."

Sid, Dad to Bill

Finding the genetic cause of mitochondrial disease

If there's no family history, but the doctor suspects mitochondrial disease based on the person's symptoms, scientists need to start searching for the gene causing the problems.

The first step to genetic testing is to collect a sample of blood, urine and/or saliva from a patient which is then sent off to a lab for testing.

Scientists begin by looking at nuclear genes known to be linked to mitochondrial disease.

They usually carry out "panel testing", which means examining all the nuclear genes currently associated with mitochondrial conditions and the panels are updated as new discoveries are made.

Mitochondrial DNA is tested separately, as faults here can also cause mitochondrial disease.

Looking at both nuclear and mitochondrial DNA helps build a clearer picture of what might be causing the condition.

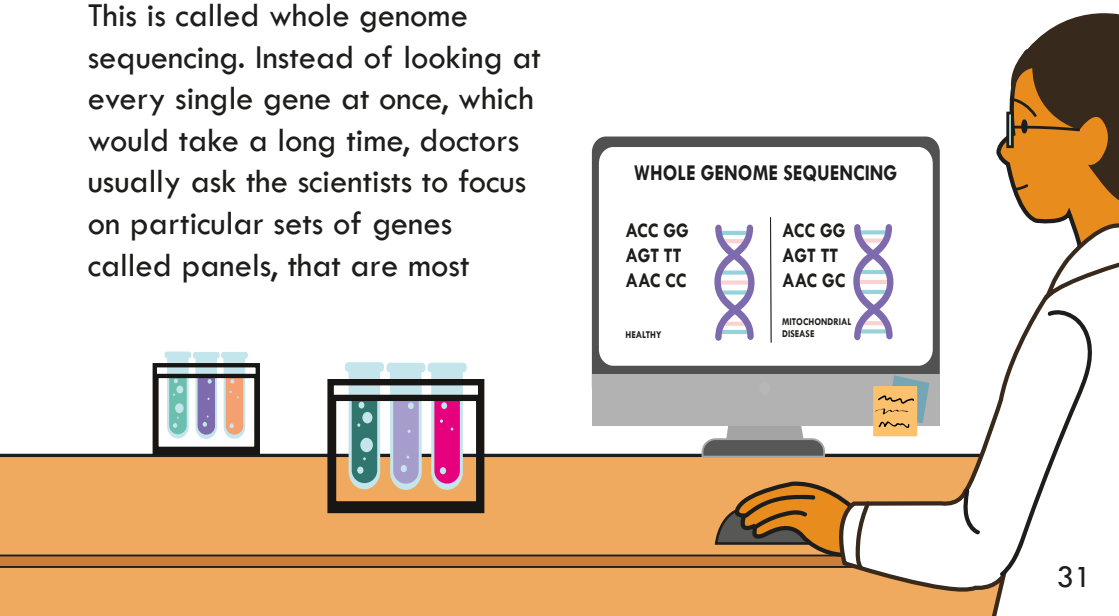
Historically, scientists could only look at genes in very small numbers at labs around the world, meaning that the diagnostic journey was long and expensive.

However, thanks to advances in technology, scientists can now read through almost all of a person's genetic code in one go.

This is called whole genome sequencing. Instead of looking at every single gene at once, which would take a long time, doctors usually ask the scientists to focus on particular sets of genes called panels, that are most likely to be linked to the condition being investigated. By comparing the person's DNA to a reference 'blueprint' of healthy human DNA, they can look for differences that might explain the cause of mitochondrial disease.

Occasionally, the panel might be quite selective and focus on only one or a small number of genes.

This only happens when testing relatives of confirmed mitochondrial disease patients.



A doctor suspects mitochondrial disease. Samples (blood, urine and/or saliva) are collected for genetic testing.

Scientists begin the search for the disease-causing mutation. Once located, they must prove it causes disease.

Genetic testing is performed. The process may take several months due to the complexities of searching across two different genomes.

Modern techniques such as whole genome sequencing allow scientists to study vast amounts of genetic data.

If a genetic cause is not found, doctors may give a clinical diagnosis based on symptoms. A muscle biopsy can pinpoint the problem and provide a biochemical diagnosis.

Undiagnosed patients may be enrolled in research projects. These use advanced techniques and analysis to find hidden or new genetic faults, and give definitive genetic diagnoses.

In children with suspected mitochondrial disease, samples are also often taken from Mum and Dad, so that scientists can compare the child's DNA to each parent.

This helps them work out whether a change in the child's genes has been inherited or has happened for the first time, which can be an important clue in identifying exactly where the genetic change has occurred.

What makes this process a little more complicated is the fact that we all have many genetic variations within our own DNA blueprint, which is what makes us unique.

Most aren't capable of causing disease, but they make the scientists' job even harder, because not only do they have to locate the exact genetic variation, they also have to prove that it's responsible for causing disease.

Sometimes additional biochemical testing is needed to check if the mitochondria has been assembled correctly and is functioning properly.

These tests help prove whether a genetic variation is responsible for the disease.

This means that laboratory testing can take several months to confirm a result which is then communicated to the patient or family.

Modern genetic techniques are using technologies that investigate our DNA in even more detail and are leading to breakthroughs in the number of people being diagnosed with mitochondrial disease, though there are still a number of patients who have been unable to be diagnosed despite extensive genetic testing.

For these patients, ongoing research projects funded by The Lily Foundation continue to use cutting-edge techniques to finally find a genetic diagnosis for patients and their families.

Signs and symptoms of mitochondrial disease

Because mitochondria are present in almost every cell in our body, people can develop a wide range of symptoms depending on which cells are affected by faulty mitochondria.

Even within members of the same family, people can be affected in completely different ways, with one person much more severely affected than another.

This is why we sometimes use the phrase 'any symptom, any organ, any age'.

"The Lily Foundation… are a breath of fresh air and have brought so many amazing people into my life. The incredible friends I have made all with a similar story to myself. I can ask questions and get answers. The Lily Foundation have become a special part of my life."

Amy, adult with mito

Genes and Syndromes

When you receive a genetic diagnosis, you may be told the name of the faulty gene that's been responsible for causing mitochondrial disease.

This is likely to be a series of letters and numbers, such as MT-ATP6, SURF1 or m.3243A>G, and these refer to the exact gene or gene location where the problem has occurred.

Some of the more common mitochondrial diseases are known by a syndrome, for example Leigh syndrome, Pearson's syndrome and Kearns-Sayre syndrome (KSS).

These groups of diseases have typically been named after the doctors who discovered them and are characterised by a particular set of symptoms.

However, it's important to note that even if you have a syndrome diagnosis, it doesn't necessarily mean that you'll develop every symptom associated with it.

There's also often a large amount of variation in symptoms and severity depending on whether a person is diagnosed in childhood or adulthood.

Mitochondrial diseases that begin to show symptoms in childhood tend to be more severe and are often diagnosed in hospital after a period of illness or an emergency admission.

Common features of mitochondrial diseases that begin in childhood may include problems with growth and development, failure to meet normal childhood milestones and the loss of previously acquired skills, as well as possible stroke-like episodes or seizures.

When the mitochondria don't function as they should, the body's energy production

becomes less efficient, leading to a build-up of lactic acid and other acids in the blood. This is known as metabolic acidosis.

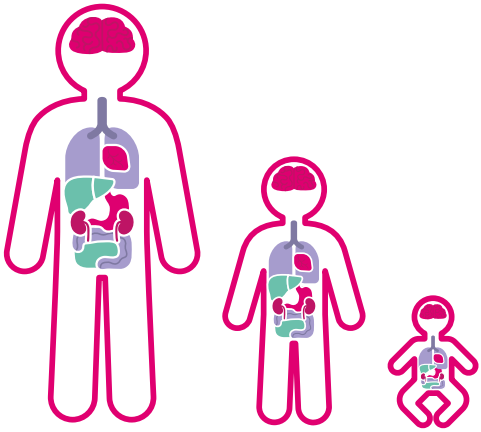
During times of illness or physical stress, this imbalance can worsen, causing decompensation, when the body can no longer keep up with its energy needs, which may make the child suddenly more tired, weak or seriously unwell.

On the other hand, adults with mitochondrial disease tend to face a longer path to diagnosis, and often present with multiple symptoms that are common amongst other conditions, such as fatigue, diabetes, hearing problems or vision loss.

It's often only when a medical professional begins to connect these seemingly unrelated symptoms that mitochondrial disease is suspected and tested for.

Sometimes, several generations of the same family are affected in this way before mitochondrial disease is eventually diagnosed.

Increased awareness and education amongst GPs and non-specialist professionals is needed to shorten the time to diagnosis for these patients.



Any Symptom
Any organ
Any age


Prof. Kostas Tokatlidis
Cathcart Chair, Institute of Molecular Cell Systems and Biology
Member

Speaking to the experts

The role of the specialist mito team

Here in the UK, we're extremely fortunate to have a network of specialist centres called the Highly Specialised Service for Rare Mitochondrial Disorders, which are commissioned by NHS England but accessible to all UK residents.

There are three centres, located in Newcastle, Oxford and London. Each is a centre of clinical and research excellence, with highly trained, specialist teams dedicated to caring for people with mitochondrial disease.

Outside of the Highly Specialised Services, there are other centres which have specialist expertise in mitochondrial disease.

These centres include Cambridge, Great Ormond Street Hospital, Manchester, Bristol, Edinburgh, Evelina London Children's Hospital and Birmingham.

All have specialist mitochondrial and metabolic teams who provide excellent care for children and adults with mitochondrial disease.

"I can't imagine how much more difficult our lives would be right now without the support and guidance you've provided to all the family."

Lisa, mum of a child affected by mito

Highly Specialised Services

There are several benefits to being seen by a specialist team.

Not only does a referral enable you to get support, care and advice from a multi-disciplinary team with specialist expertise, but it also allows you to access clinical trials and research opportunities that help us understand more about mitochondrial disease and develop treatments for the future.

In order to be accepted as a patient, a referral must come from a medical professional who has either already confirmed the diagnosis of mitochondrial disease or strongly suspects that this is the case.

It's important to remember that specialist centres for mitochondrial disease are unable to provide all of your care. You may only attend a clinic appointment once every 6 months to a year, and you may live a long way away from your specialist centre.

That's why it's really important to maintain a good relationship with your GP and local hospital team.

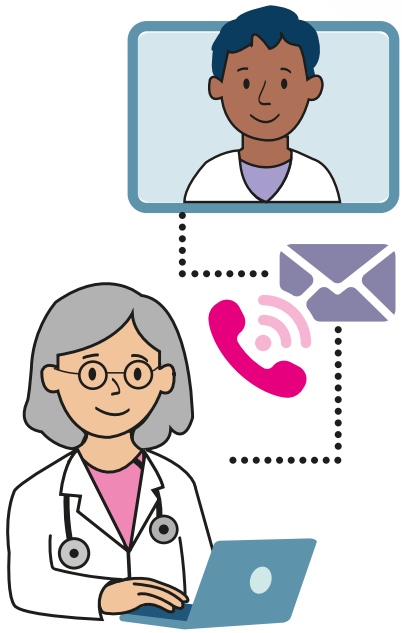
We understand that this can sometimes be challenging due to limited knowledge and awareness of many rare mitochondrial diseases; however, it's in your best interests to be able to access care and treatment as soon as possible from a medical team who are familiar to you and can assess you or your child face-to-face.

The specialist teams across the country are always willing to provide support and advice to local hospital teams, particularly in times of illness or before planned procedures.

Many centres offer medical alert cards which provide the

numbers for professionals to contact them during normal work hours and in an emergency.

They're happy to discuss care plans and special considerations for mitochondrial disease patients with local medical teams, and they've also developed national guidelines on various topics including anaesthesia and surgical considerations, cardiology, diabetes, epilepsy and physiotherapy.



These are publicly available at https://mitochondrialdisease.nhs.uk/professional-area/care-guidlines/.

Please note that these guidelines are not intended for patients, though you may wish to pass them on to your local teams.

If you're a patient who's received a medical alert card from your specialist, please always carry this with you in case you need it.

It can also be a good idea to keep your last clinic letter handy, as this document will include valuable medical information that you can pass to your local team.

There are various charity-driven initiatives, such as patient passports and medical alert bracelets, that patients can use to provide more information about themselves. Find out more in the 'Further Support' section of our website.

The role of the local team

Due to the nature of mitochondrial disease, it's likely that you may need to have a range of professionals involved in your or your child's care.

Though there are very few treatments for mitochondrial disease, screening and surveillance is very important to help ensure that you stay as healthy as possible.

Depending on your own individual circumstances, you may need to see a range of other hospital specialists who can manage your symptoms properly, such as cardiology to screen for any heart problems, or endocrinology to manage mitochondrial diabetes.

We understand that it can feel overwhelming to attend so many different appointments, particularly whilst also managing symptoms such as fatigue, which can make attending clinics challenging.

Within your local care team, you may also meet professionals out in the community including a physiotherapist, occupational therapist, social worker, speech and language therapist, dietitian and community nursing teams.

Your GP or specialist doctor can help to signpost you to the appropriate referrals.

If you're the parent/carer of a child, you may also require help to access specialist support at school, and it's a sensible idea to build a good relationship with your child's teacher or SENCO to help put provisions in place to ensure that your child can get the best experience whilst in school.

Medicines guidance

People with mitochondrial disease often have questions about whether it's safe to take certain prescribed medications, particularly if these haven't been prescribed by a specialist in mitochondrial medicine.

To address these concerns, specialists from across the world developed guidelines about which drugs are safe to take and which should be used with caution in mitochondrial disease.

The list is available for reference on our website, at:

https://www.thelilyfoundation.org.uk/affected-by-mito/all-about-mito/drug-info/.

There are very few drugs that absolutely should be avoided in a particular type of mitochondrial disease.

Sodium Valproate should not be used in patients with POLG-related disease due to the specific risk of liver failure.

In addition, aminoglycoside antibiotics such as Gentamicin should be avoided in patients with m.1555A>G due to a very high risk of causing deafness.

It's important to remember that all drugs can have side effects, even in the general population, so if you have any concerns, we encourage you to discuss these with your doctor or specialist.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Vitamins and supplements

Depending on your type of mitochondrial disease, your specialist may prescribe supplements to help ease symptoms such as fatigue.

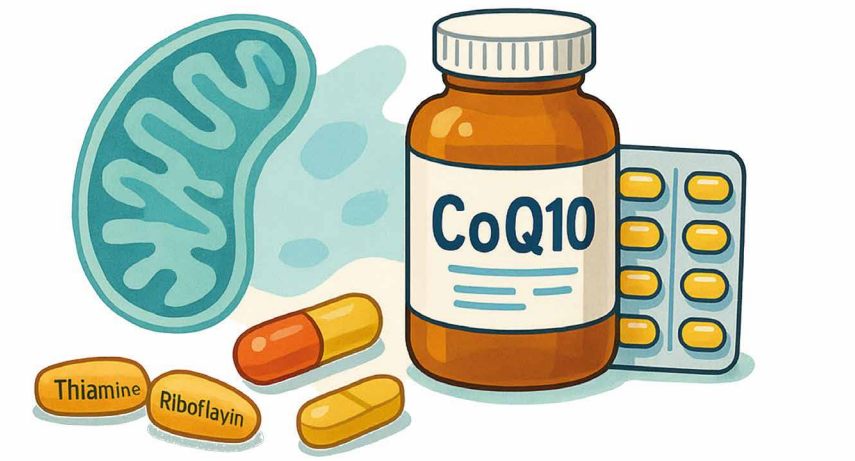
The most common is Co-Enzyme Q10, though there are also certain types of mitochondrial disease where others including Thiamine and Riboflavin may also be prescribed.

Often, specialists will start newly diagnosed patients with a course of Co-Enzyme Q10 on a trial basis for a short period, usually around 3 months.

If the patient or family feel that they've noticed a benefit, this will be continued, with other vitamins and supplements trialled as needed.

Due to a lack of clinical trials, concrete evidence about the benefits of vitamins and supplements is limited and most reports are anecdotal.

Some people don't notice any difference after trialling vitamins and supplements, while others report feeling real benefits.

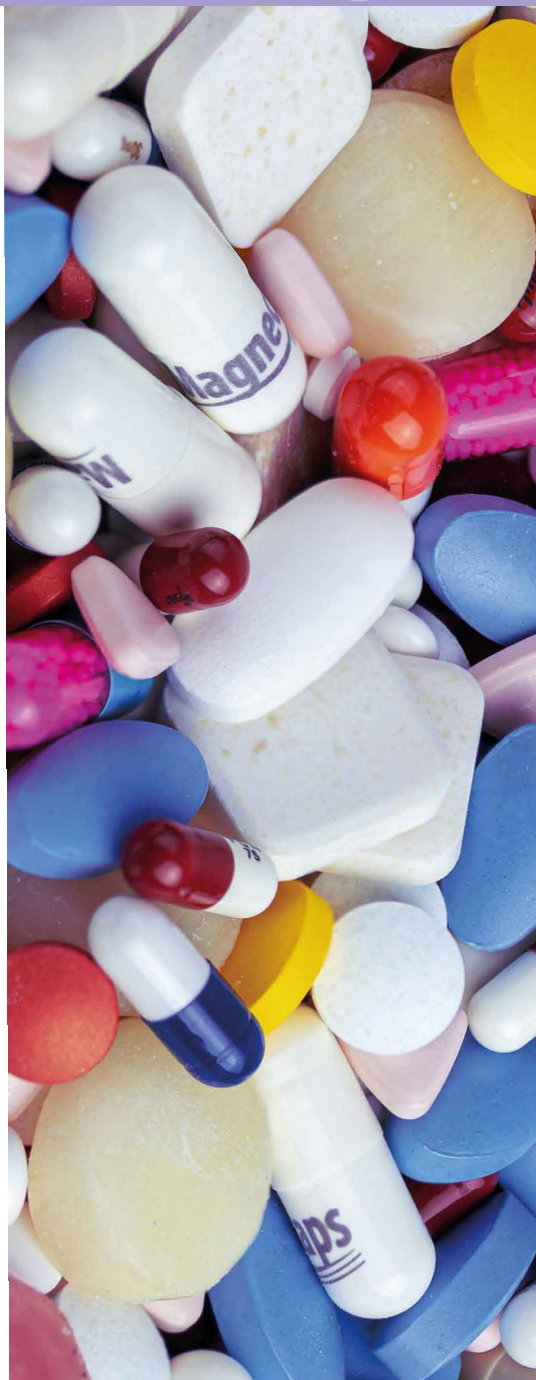


In other countries including the US, people with mitochondrial disease are more likely to be prescribed the 'mito cocktail' by their doctor, which is a combination of these vitamins and supplements prescribed together.

The use of the mito cocktail is much less common practice in Europe, where doctors are more likely to prescribe one at a time to assess each for clinical benefit.

There are other multivitamins that you can buy as over-the-counter medications to help maintain overall health.

These are used widely by the general population and are generally considered to be safe, though it's worth mentioning during your clinic appointment if you're taking any of these.



Clinical trials and research studies

Clinical trials and research studies are a vital part of the journey towards finding treatments for mitochondrial disease.

They help scientists and research teams understand more about what's causing disease, and test out possible new treatments to find out whether they're safe and effective.

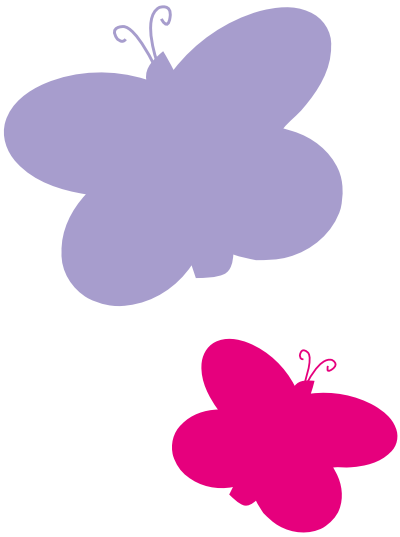
Clinical trials can also help rule out treatments that don't work, meaning that researchers can then start to look at new approaches.

Whilst clinical trials for mitochondrial diseases are still relatively limited, there's a growing interest in the role that mitochondria may play in overall health.

Because mitochondria play such a crucial role within all the cells in our body, they've been linked to more common illnesses such as cancer and dementia. It's hoped that studying mitochondrial diseases may also unlock treatments and cures for these illnesses too.

Clinical trials can vary in length and may require different demands on the person or family taking part.

Some studies are observational, meaning there's no active treatment being tested, whilst others may be testing a new medication or device.



As our understanding of genetic diseases develops, we're likely to see a rise in the number of clinical trials testing advanced therapies, such as gene or stem cell therapy.

It's important to understand the potential risks and benefits of any clinical trial before deciding whether or not to take part.

The decision to take part in any form of research is deeply personal and should be made voluntarily by the patient and family after being given clear, balanced information to make an informed choice.

If you're interested in clinical trials, it's a good idea to request a referral to one of the specialist teams for mitochondrial disease if you're not already under their care.

Once there, you can request to be added to their research database to be contacted for any future trials you may be eligible for.

You can also check the UK trials and research studies pages of The Lily Foundation website to find out which trials are currently recruiting for participants.


Donna Bagatti
Lily Volunteer

The importance of peer support

The role of patient organisations

Here at The Lily Foundation, we understand the strength and power of community in helping to support patients and families facing a diagnosis of mitochondrial disease.

Receiving the diagnosis can not only be devastating but also lead to feelings of uncertainty and isolation.

Because of the rarity and complexity of mitochondrial disease, as well as a lack of awareness of the condition, it can be exhausting to have to constantly explain your diagnosis to family, friends and loved ones.

Often, the symptoms of mitochondrial disease aren't clearly visible, so patients and family members feel misunderstood, ignored or stigmatised because they're unable to live the life they wished for.

This is where The Lily Foundation can help.

"I honestly don't know where we'd be without The Lily Foundation now. We'd be a very different couple and a very different family. The friends we've made are incredible and so much positivity has come out of our involvement with the charity."

Kerry, Mum of Bill

Ways in which we can support you

Amongst the many services we offer, we can provide a safe, confidential space to speak to others about how you're feeling with someone who understands and who may have walked a similar journey to yourself.

We appreciate the value of being able to ask questions and get advice from others who have gone before you, so we offer one-to-one and peer support over the phone, via email and on video calls.

We have private closed Facebook groups for families and adults, which are carefully moderated, and we offer weekly community catch-ups through our virtual coffee morning.

By far the largest events in the calendar are our annual residential weekends for our families, young people and adults affected by mito.

These much-needed occasions provide a real sense of community and help break down the isolation faced by many patients and families with mitochondrial disease.

In addition, we provide education about mitochondrial disease to patients, the public and non-specialist professionals, including speaking at conferences, running online webinars and attending Patient Information Days.

We can help people to navigate local support services and signpost to specialists who can assist with the many challenges of living with mitochondrial disease.

We also have access to a free service that offers counselling sessions to anyone affected by mitochondrial disease via our partnership with Rareminds, a non-profit organisation that specialises in mental health support for people living with rare diseases.

We're passionate about raising the profile of mitochondrial disease and putting patient voices at the heart of everything we do.

Our programme of patient engagement provides people affected by mitochondrial disease with a platform to have their voices heard, offering various opportunities to share their personal stories and have their say in shaping clinical services and the research of tomorrow.

Feel free to contact us...

Finally, we fund medical research, because we know that for patients and families affected by mitochondrial disease, their number one priority is to find a cure.

That's our priority too.

Over the years we've heard many people tell us that being part of The Lily Foundation feels like being part of a family, and that at the darkest time, we've helped them to face their diagnosis and find hope.

We'd love to support you too. Please don't hesitate to contact us to find out more about how we can help.

Contact Liz, our CEO and Founder, on 07947 257 247

liz@thelilyfoundation.org.uk

We hope to hear from you soon.


Fighting mitochondrial disease.
Lily
Fighting for hope.
thelilyfoundation.org.uk
NK70 ZKH

You're not alone

Living with mitochondrial disease can feel isolating, but through The Lily Foundation you can stay connected to a community that understands.

By signing up to our newsletter, you'll receive important updates on our support services, advocacy work and research breakthroughs, as well as the latest news about Lily events, fundraising campaigns, personal stories and more. Scan the QR code below to join our community.

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