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What is mitochondrial disease and what causes it?

Mitochondrial disease (or mito) is a debilitating genetic disorder that robs the body's cells of energy, causing multiple organ dysfunction or failure and potentially death. Most patients have a genetic mistake (mutation) in the mitochondrial or nuclear DNA. The condition can be inherited from the mother, the father or both parents, or can arise as a spontaneous genetic mistake at conception.

What are mitochondria?

Mitochondria are the energy source in almost every body cell. Often called the cells' powerhouses or generators, mitochondria transform food to produce 90 per cent of the energy needed by the human body to function, sustain life and support growth. Mitochondria are most plentiful in tissues that require a lot of energy to function; the disease therefore causes most damage to the cells of the brain, muscles, heart, liver, inner ear and eye.

How are sufferers affected?

Depending on which parts of their bodies are affected and to what degree, people with mitochondrial disease can:
- lose their sight or hearing
- suffer muscle weakness and pain
- be unable to walk, eat, swallow or talk normally
- have strokes or seizures
- develop liver disease or diabetes
- suffer heart, respiratory or digestive problems
- experience developmental delays or intellectual disability.

Needing to stay in bed to rest and recharge is a common outward symptom of mitochondrial disease. Inside the body, it's much more serious and complex: mitochondrial disease may literally cause any symptom in any organ at any age.

Who is affected by mitochondrial disease?

Mitochondrial disease can affect both children and adults; due to its genetic basis, the disease often affects multiple family members. Adult onset is becoming more commonly recognised. In many cases, the impaired mitochondrial load (cell injury and cell death) increases with age, until organ systems begin to fail and symptoms develop.

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**How common is mitochondrial disease?**

Until the 1990s, mitochondrial disease was thought to be rare (1 in 20,000 people), but it is now recognised as the most common subgroup of inherited metabolic disorders. Recent research shows up to 20 children born in Australia each week (1 in 250 people) may carry genetic mutations that put them at risk of developing mild forms of mitochondrial disease or other related conditions such as diabetes or deafness during their lifetimes. This means 90,000 Australians may be affected by mitochondrial disease. Many of these people are symptomatic but undiagnosed or misdiagnosed, some are not yet symptomatic, and others are unknowingly at risk of passing the disease on to their children.

In addition, one Australian child born each week – or 50 children every year - will develop a severe or life-threatening form of mitochondrial disease (1 in 5000 people), making it the second most commonly diagnosed serious genetic disease after cystic fibrosis, which has an incidence of around 1 in 3500 people.

**How is mitochondrial disease diagnosed and treated? Is there a cure?**

Mitochondrial disease is a complex condition that is difficult to diagnose due to the widespread range, type and severity of symptoms and its varying onset and impact on patients’ lives (from none to severe). Multiple tests may be required to confirm mitochondrial disease, including genetic tests, muscle biopsies or brain scans (depending on the type of disease suspected).

There are currently very few effective treatments and as yet no cure for mitochondrial disease. It impacts differently on every patient, so doctors can’t predict the progression of the disease or symptoms, or the outcome for patients.

**Links with ageing and major diseases**

Whereas people with mitochondrial disease have a genetic mutation that predisposes their mitochondria to fail early, mitochondrial dysfunction is thought to be one of the major factors contributing to ageing and the reason why humans have a finite lifespan. Over a lifetime, our mitochondria slowly suffer inevitable damage from environmental and lifestyle factors and become less effective at producing the energy our organs need to function properly.

Researchers increasingly believe mitochondrial dysfunction may be a significant factor in a wide range of major diseases – particularly chronic degenerative disorders and those associated with ageing – including:

- Parkinson disease
- Alzheimer disease
- Huntington disease
- motor neurone disease / amyotrophic lateral sclerosis (ALS)
- cardiovascular disease
- diabetes
- cancer, particularly solid tumours and tumour metastasis (spread to other organs).

Research into mitochondrial medicine therefore offers hope not only to people with primary mitochondrial disease (due to a genetic mutation), but also to the millions suffering from other major diseases commonly associated with ageing.

Improvements in mitochondrial medicine may eventually provide the key to better health and quality of life in old age for all.
Why haven’t we heard much about mitochondrial disease before?

Mitochondrial medicine is a newly established and rapidly evolving field thanks to major advances in our understanding of genetics. It was not until 1988 when mutations in mitochondrial DNA were discovered to cause disease, and not until 1995 when nuclear gene mutations were also found to cause mitochondrial disease. Since then, more than 100 clinical syndromes and disorders have been recognised as coming under the category of mitochondrial disease.

The Australian Mitochondrial Disease Foundation (AMDF)

The Australian Mitochondrial Disease Foundation was set up in 2009 by family members, friends and doctors of sufferers to fund essential research into the diagnosis, treatment and cure of mitochondrial disorders, and to support affected individuals and families.

The AMDF has already funded three PhD research projects and a fourth is in the pipeline. It has helped establish an Australia-wide mitochondrial patient database, and has also funded priority access to a new Next-Generation DNA Sequencing Facility at Royal Perth Hospital that will enable faster, less expensive and more accurate diagnoses of mitochondrial disease.


World Stay in Bed Day on Sunday 23 September 2012 is the AMDF’s major fundraising activity, conceived because being forced to stay in bed to rest and recharge is a common outward symptom of mitochondrial disease (and because we all enjoy a sleep-in once in a while).

To help sufferers get out of bed, Australians raise money by being sponsored to stay in bed until midday on 23 September via www.stayinbedday.org, hosting a pyjama party or ‘bed-in’, or making a donation. The event also features the first virtual worldwide bed on the worldwide web, via facebook.com/stayinbedday and twitter.com/stayinbedday (tweet using #stayinbedday). All funds raised go towards vital research into treatments and a cure.

A successful Australian initiative that began in 2009, Stay in Bed Day took our laid-back, generous attitude to the world for the first time in 2011, in collaboration with NZmito, the United Mitochondrial Disease Foundation (US), The Lily Foundation (UK), and Mito Canada.

The week before World Stay in Bed Day is Global Mitochondrial Disease Awareness Week (16-22 September 2012; www.gmdaw.org). It includes a variety of education and information sessions, fundraising activities, and support and advocacy initiatives.

This fact sheet has been reviewed by the AMDF’s Scientific and Medical Advisory Panel. It has been prepared for general information only and should not be relied on for decisions regarding medical care.

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