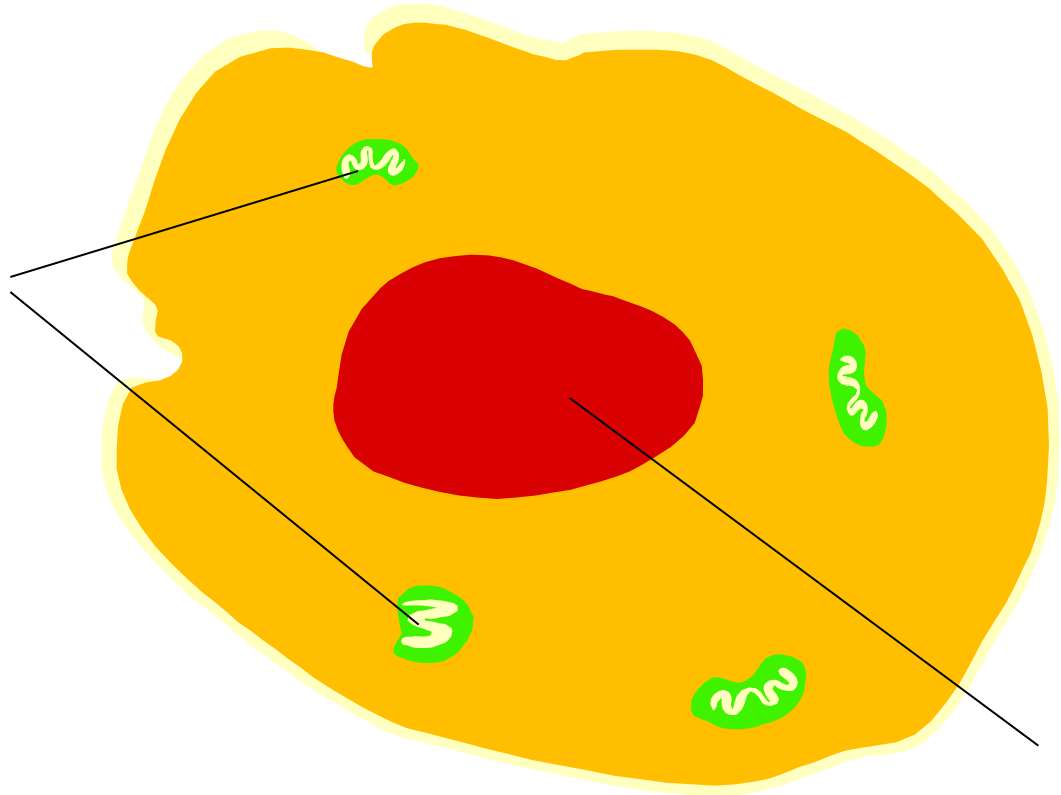
Mitochondrial Disease

What is that???

And where is that good GP to help me??

The Cells – are the basic building blocks of our body



Mitochondria

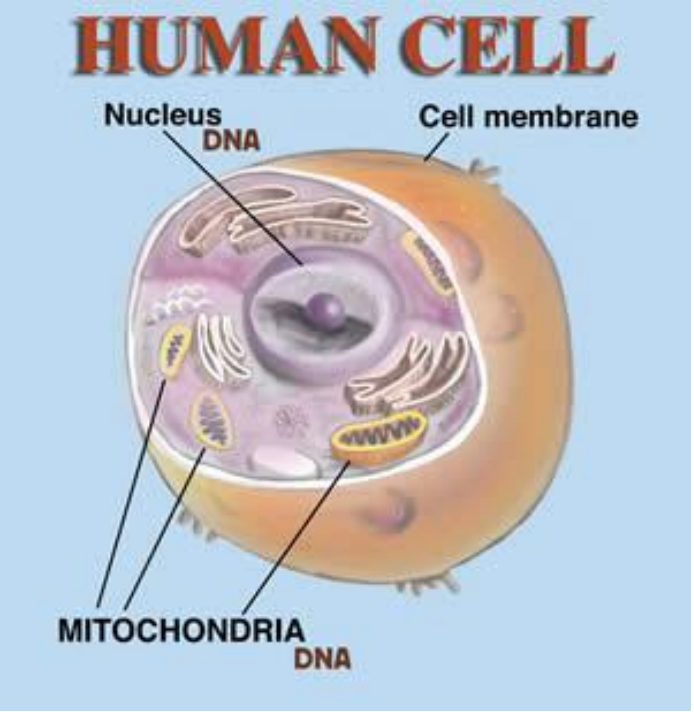
Like the batteries of the cell

Nucleus

Like the brains of the cell

What are mitochondria?

- it takes about 1400 genes to make a single mitochondria, and these are spread between the DNA on the mitochondria or the nucleus….so all sorts of inheritance which means **it is GENETIC** !!!
- So far we know of at least 200 different "subgroups" of mito disease!
- Mitochondria provide 90% of the energy needed by our body to LIVE
- the organs that need lots of energy to work, require the most mitochondria in their cells
- so those cells in the brain, muscles, heart, liver, kidneys, bowels, ears and eyes, are the most often affected



What is Mitochondrial disease then?

"Mitochondrial Disease" = mitochondria not working properly

MITO disease

Depleting lifes energy.

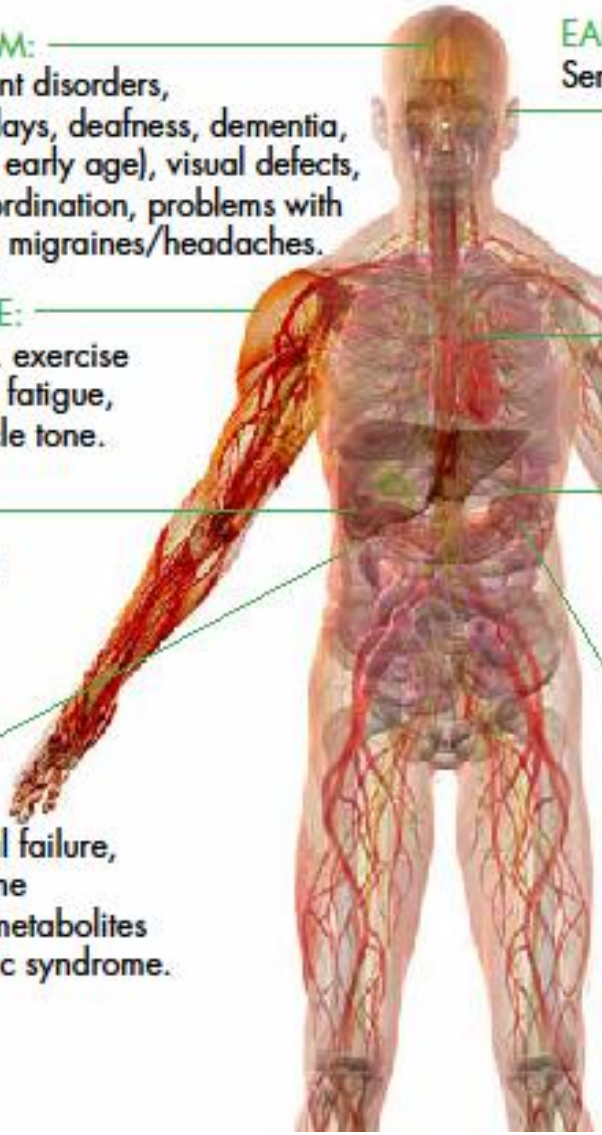
1 in 200 people carry a mitochondrial genetic defect …with a range of presentations……..nothing to 1 in 5000 developing severe Mito disease…..

this means that ***every week*** in Australia 20 children are born with a mito gene defect, and one of them will develop severe illness of whom 50% will start in childhood and most won't see adulthood…..on average every fulltime GP has 8-10 patients with a mito gene defect and every 2nd one has a serious patient!!

SECOND most common serious genetic disorder behind Cystic Fibrosis (1 in 3500)

It is generally progressive but at different rates, when mitochondria fail, cells fail, then whole organs begin to fail too, the life of the person is severely compromised and hence it is often fatal, especially in children……e.g. the poorer the battery quality, the quicker they give up

Symptoms of Mitochondrial Disease?



NERVOUS SYSTEM:
Seizures, movement disorders, developmental delays, deafness, dementia, stroke (often at an early age), visual defects, poor balance/coordination, problems with peripheral nerves, migraines/headaches.

SKELETAL MUSCLE:
Muscle weakness, exercise intolerance, pain, fatigue, cramps, low muscle tone.

LIVER:
Unexplained liver failure.

KIDNEYS:
Unexplained renal failure, Fanconi's syndrome (loss of essential metabolites in urine), nephrotic syndrome.

EARS:
Sensorineural deafness (may be intermittent).

EYES:
Drooping eyelids (ptosis), inability to move eyes (external ophthalmoplegia), blindness (retinitis pigmentosa, and optic atrophy).

HEART:
Cardiomyopathy (large heart), conduction abnormalities.

DIGESTIVE TRACT:
Difficulty swallowing, reflux, vomiting, feeling of being full, chronic constipation, diarrhoea, unexplained intestinal obstruction, irritable bowel type symptoms.

ENDOCRINE:
Diabetes hypothyroidism, and excess body hair.

It is very hard to find who has it because “it can affect any organ, have any symptom, and at any age”!!

So it is often missed for years!!

Therefore it has been very aptly named……

“THE NOTORIOUS MASQUERADER”

Diagnosis?

Like building up a jigsaw puzzle

- history (yourself and family)
- symptoms
- basic tests
- specialised mitochondrial function tests
- genetic tests (complicated, can take a long time with no result, like a needle in hay stack)…the final piece but not 100% needed

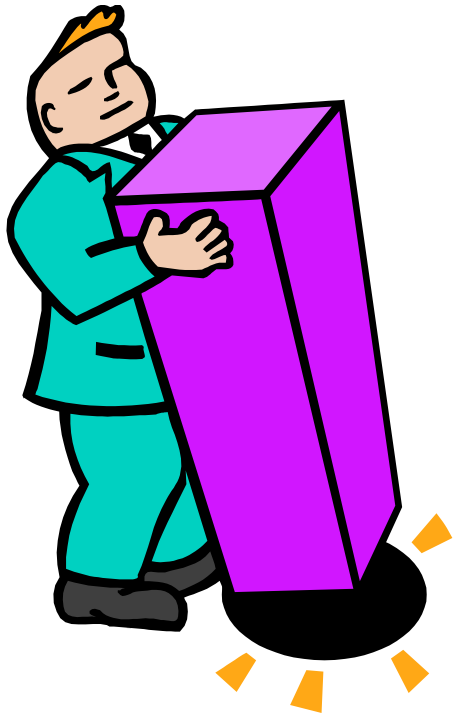
So why have so many not heard of it yet especially in the medical profession?

- complicated genetics, no one pattern even in families
- relatively new illness, on a major learning curve still
- mimics or masquerades as other illnesses
- difficult to diagnose…clinically, biochemically and genetically
- no clear classification system
- very slow and insidious onset in the majority…. especially in adults

When should my GP think of it ?

1) Three or more organ systems are involved

(GP can see this better than specialist)



2) A ‘common disease’ has atypical features that set it apart from the pack

3) Recurrent setbacks/flare ups in a chronic disease occurs with infections

Treatment?

There is currently **no cure** for mitochondrial disease… so what is the POINT??

BUT Treatment focuses on a TEAM APPROACH for……

Alleviating symptoms, e.g. surgical correction of droopy eyelids and weak eye muscles, hearing aids, medications (especially for seizures), bowels, etc

Regular monitoring of the systems and earlier management (e.g. regular echoes)

Optimising the mitochondrial environment = the 'mitochondrial cocktail'
There is no 'standard' cocktail though.

Keep the energy equation balanced = avoid upsetting the balance
thru appropriate **lifestyle changes**

"Benefits" for the family, helping those who may have similar symptoms, genetics

"Benefits" for self, a diagnosis can lead to better access to services/resources

??? Palliation

Goals of “Treatment”

Brain
– reduce seizures
– improve attention and concentration
– improve intellectual functioning
– prevent headaches
– prevent strokes
– improve motor control

Muscle
– improve strength
– lessen pain
– lessen fatigue
– reverse cardiomyopathy

Liver
– improve function, avoid “toxins”

Nerve
– improve autonomic function
– lessen pain
– improve nerve conduction

GI
– improve gastric and intestinal motility

Eyes
– prevent further retinitis or optic atrophy

Ears
– prevent further hearing loss

Systemic
– encourage growth and so prevent **failure to thrive**

Monitor for complications ……REVIEW REGULARLY, a long path with GP!

Monitoring = “25,000km check up”

- Blood for full biochemistry, HBA1C, TSH, B12 and FBC/film
- Serum lactate, CK, carnitine levels, etc
- Neuroradiology (CT scan)
- Electrophysiology (EEG, NCS, EMG, etc)
- Audiology assessment
- Ophthalmological review
- Gastroenterological review
- Cardiological review (ECG, holter, echo)
- Allied health team assessments (OT, speech pathologist, physiotherapist)

“Pharmacological” treatment

- Coenzyme Q10, Idebenone

- Vit K, C, E, carnitine, creatine, thiamine, riboflavin, magnesium orotate
- Cocktails – vary between subgroups and specialists/clinics

Prophylactic treatment

ENERGY IN = ENERGY OUT

Avoid upsetting the balance (GP can help heaps here!)

- **Energy In** - mitochondrial cocktail, diet including adequate/regular meals and snacks, avoid fasting, adequate sleep and rest, regular exercise program

- **Energy Out** – prevent infections, avoid EXCESS stresses such as exertional/emotional, avoid toxins, adequate rest, avoid big temperature changes, care with any operations

Palliation?

Symptom Control

• common mito symptoms include seizures (esp. uncontrolled), autonomic instability, gut dysmotility, constipation, pain/cramps, anxiety/dementia, aspiration, sepsis, weakness, end organ failures (e.g. cardiac, renal), blindness, deafness

Physical Support

• home modification, beds, suction, oxygen, feeding supplies, lifts, bath, home nursing and/or respite

Psychosocial Support

• treatment, advanced care planning, discussing anticipated course, patient/family values, end-of-life preferences, realistic goals for life, maintenance of functionality – reframe/rethink

Emotional Support

• caregivers, siblings of affected children, parents, grandparents and spouses

Spiritual Support

• spiritual care provider

Family Considerations

• genetic counselling, coordination of care and case management, finances (power of attorney)

End-of-Life Care

• early referral to palliative care, helping family with memory making (e.g. scrapbooking)

amdf+

AUSTRALIAN MITOCHONDRIAL DISEASE FOUNDATION

our mission

research + support + educate

to foster research towards a cure for mitochondrial disease

to provide support for sufferers of mitochondrial disease & their families

to promote education regarding mitochondrial disease for the general public & the medical profession

THANK YOU

“An observant parent’s evidence may be disproved but should never be ignored”

—Lancet 1:688, 1951, Anonymous