MAYBE IT'S MITO

1 any symptom

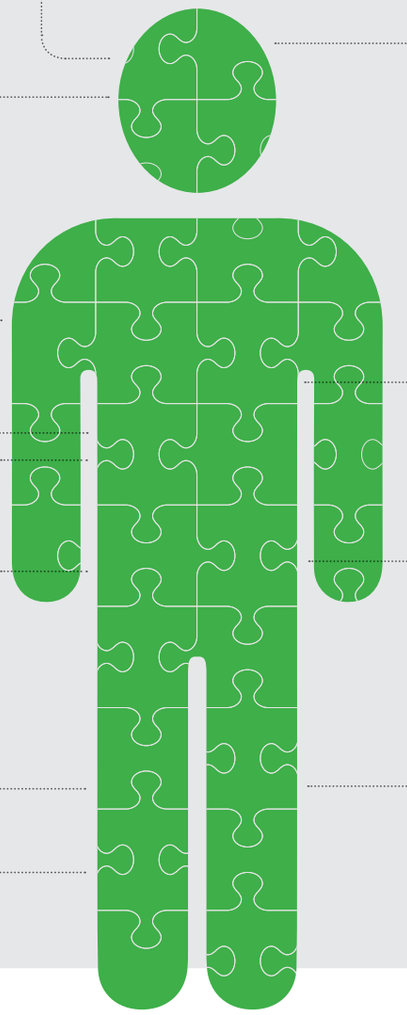
A 'common' disease presents with 'atypical' features

Diagnosing primary mitochondrial disease or 'mito' can be challenging due to the wide variety of symptoms and sub-groups.[1] Many experts refer to it as the 'notorious masquerader' because it mimics so many other different illnesses.

2 any organ

Three or more organ systems are involved and/or any 'red flag' symptoms are present[1,2]

EYES
Visual loss/blindness, optic atrophy, disorders of extra-ocular muscles, ptosis, retinal degeneration with signs of night blindness, colour-vision deficits, pigmentary retinal changes such as retinitis pigmentosa or 'salt and pepper' retinopathy.



EARS
Hearing loss and deafness, sensorineural hearing loss.

BRAIN & NERVES
Developmental delays, mental retardation/regression, focal neurological deficits, dementia, seizures, coma, neuro-psychiatric disturbances, atypical cerebral palsy, myoclonus, movement disorders, ataxia, migraines, strokes, weakness (which may be intermittent), neuropathies, absent reflexes, fainting, absent or excessive sweating resulting in temperature regulation problems.

MUSCLES
Weakness, hypotonia, cramping, muscle pain, recurrent rhabdomyolysis.

HEART
Conduction defects (e.g., heart blocks, WPW), cardiomyopathy.

LIVER
Hypoglycaemia, unexplained liver failure, valproate-induced liver failure.

PANCREAS
Diabetes and exocrine pancreatic failure (inability to make digestive enzymes).

KIDNEYS
Proximal renal tubular wasting resulting in loss of protein, magnesium, phosphorous, calcium and other electrolytes, aminoaciduria, nephrotic syndrome.

DIGESTIVE
Gastro-oesophageal reflux, delayed gastric emptying, constipation, pseudo-obstruction, chronic or recurrent vomiting.

SKIN
Symmetrical lipomatosis.

SYSTEMIC – WHOLE BODY
Exercise intolerance not in proportion to weakness, fatigue, short statue, respiratory problems including intermittent air hunger, hypersensitive to general anaesthetics.

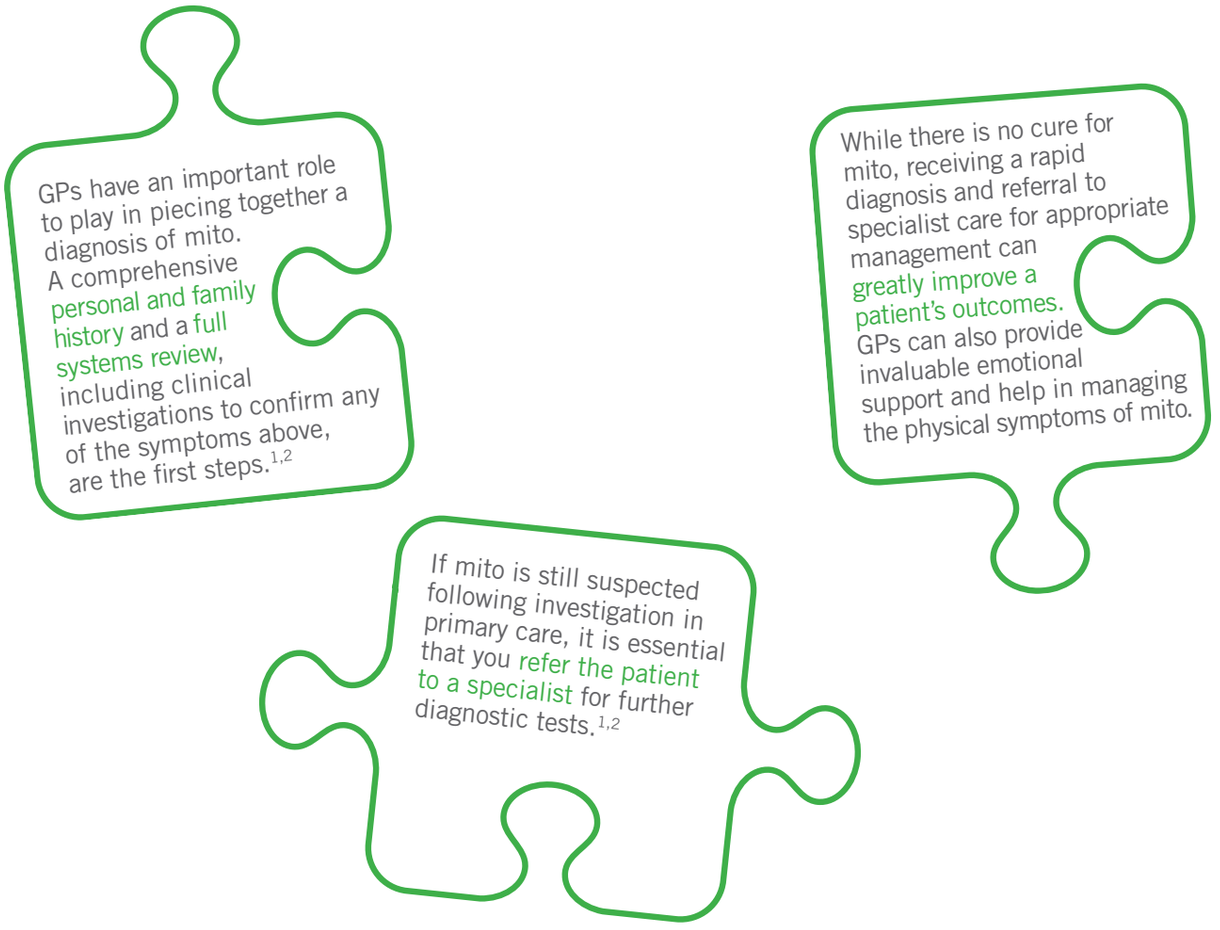
BLOOD
Sideroblastic anaemia.

3 any age

Recurrent setbacks/flare ups in a chronic disease occur with infections

Mito is the most common inherited form of metabolic disease. It is caused by genetic mutations that disrupt the production of energy by mitochondria.[3] As a result, people with mito may experience profound and prolonged fatigue, as well as worsening of existing symptoms, in response to infections.[4]

Piecing together a diagnosis of mito



Visit www.mito.org.au for more information or to find a specialist.

References: 1. Haas RH *et al. Pediatrics* 2007;120(6):1326–33. **2.** Naviaux RK. A Primary Care Physician's Guide. the Spectrum of Mitochondrial Disease. **3.** Alston CL *et al. J Pathol* 2017;241(2):236–50. **4.** Mitoaction. Immune function and mitochondrial disease. Available at: www.mitoaction.org/blog/immune-function-and-mitochondrial-disease. Accessed: 26 June 2018.

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