

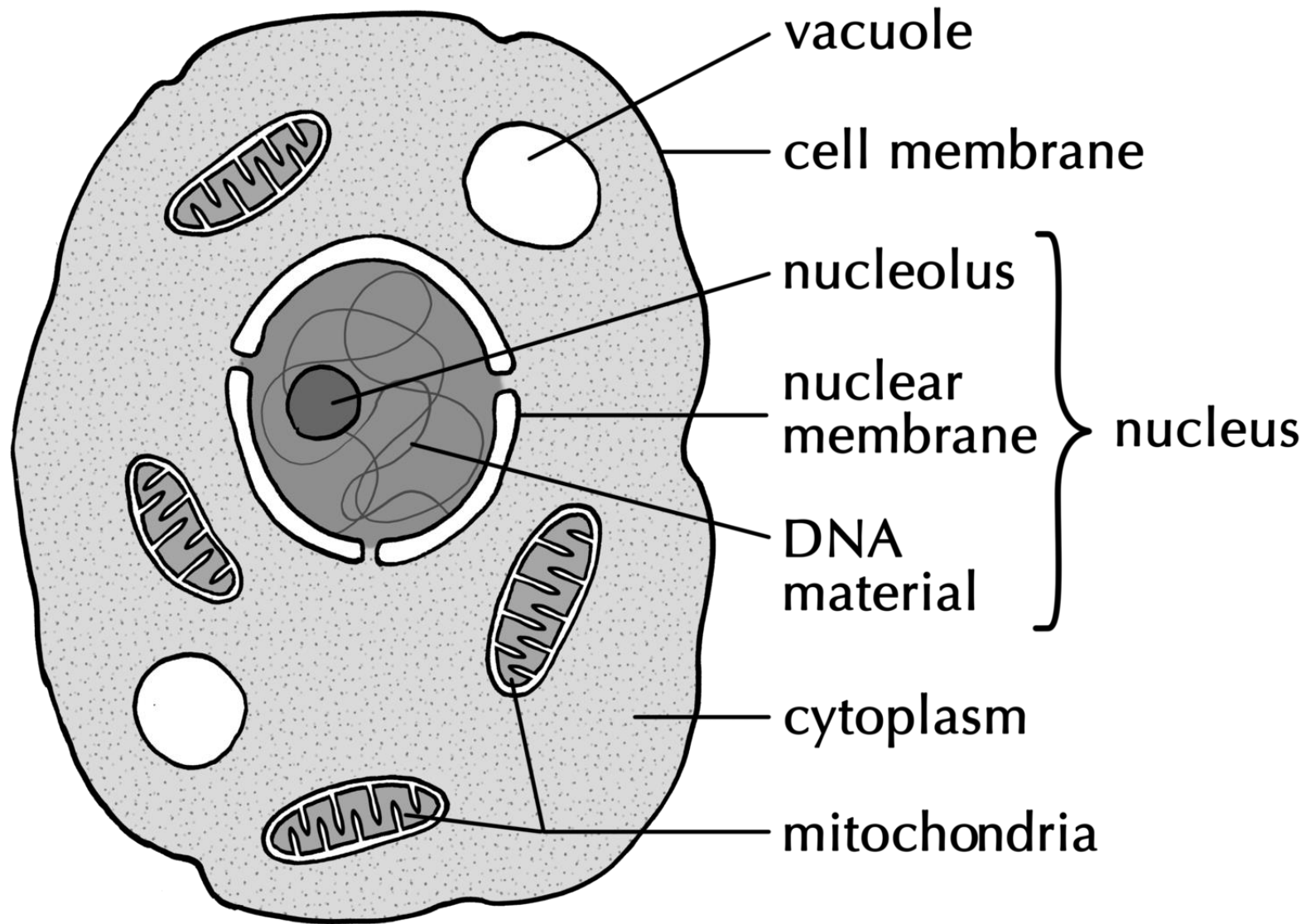
# Tests in patients with mitochondrial disease

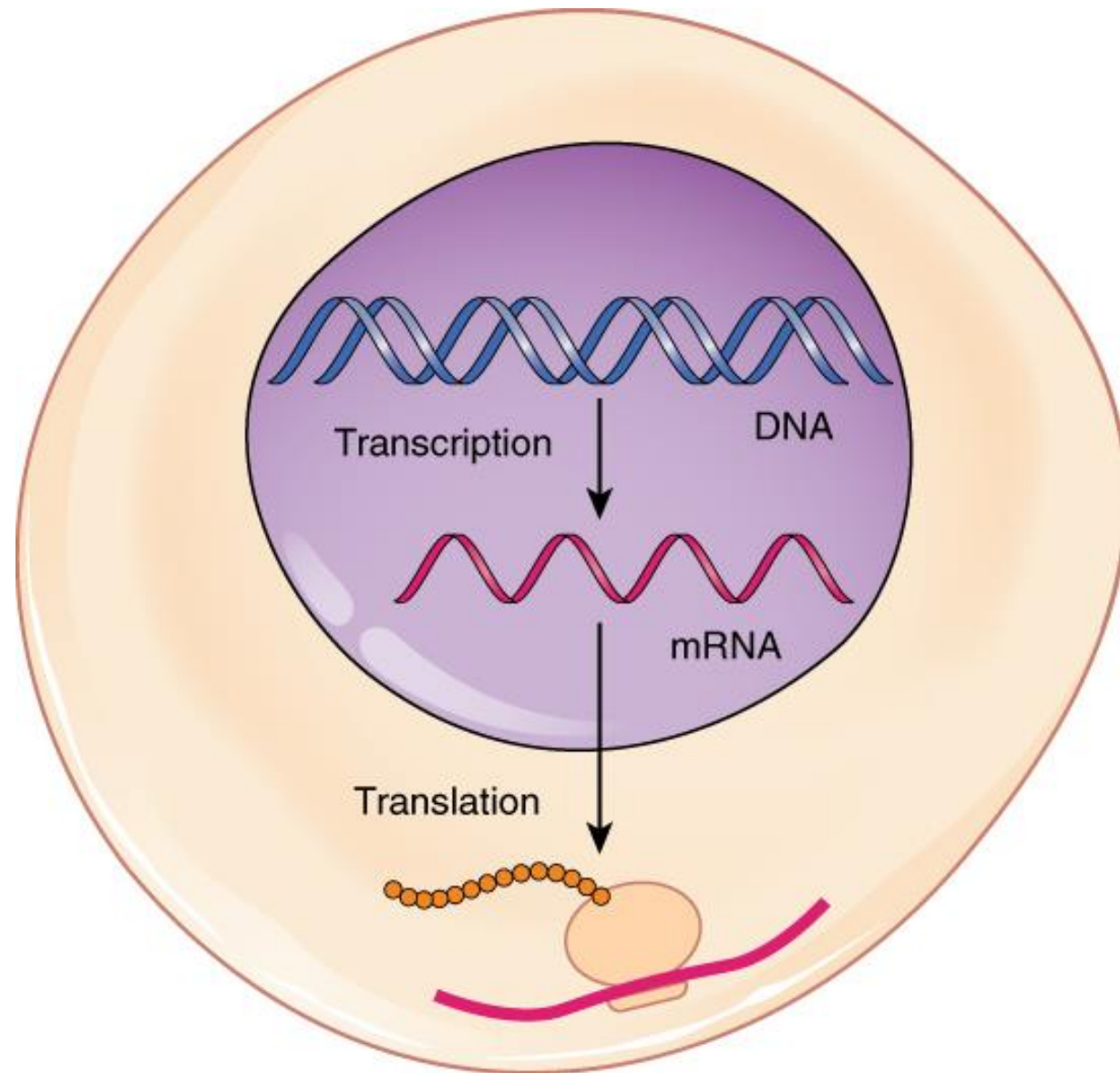
Dr Nishan Guha

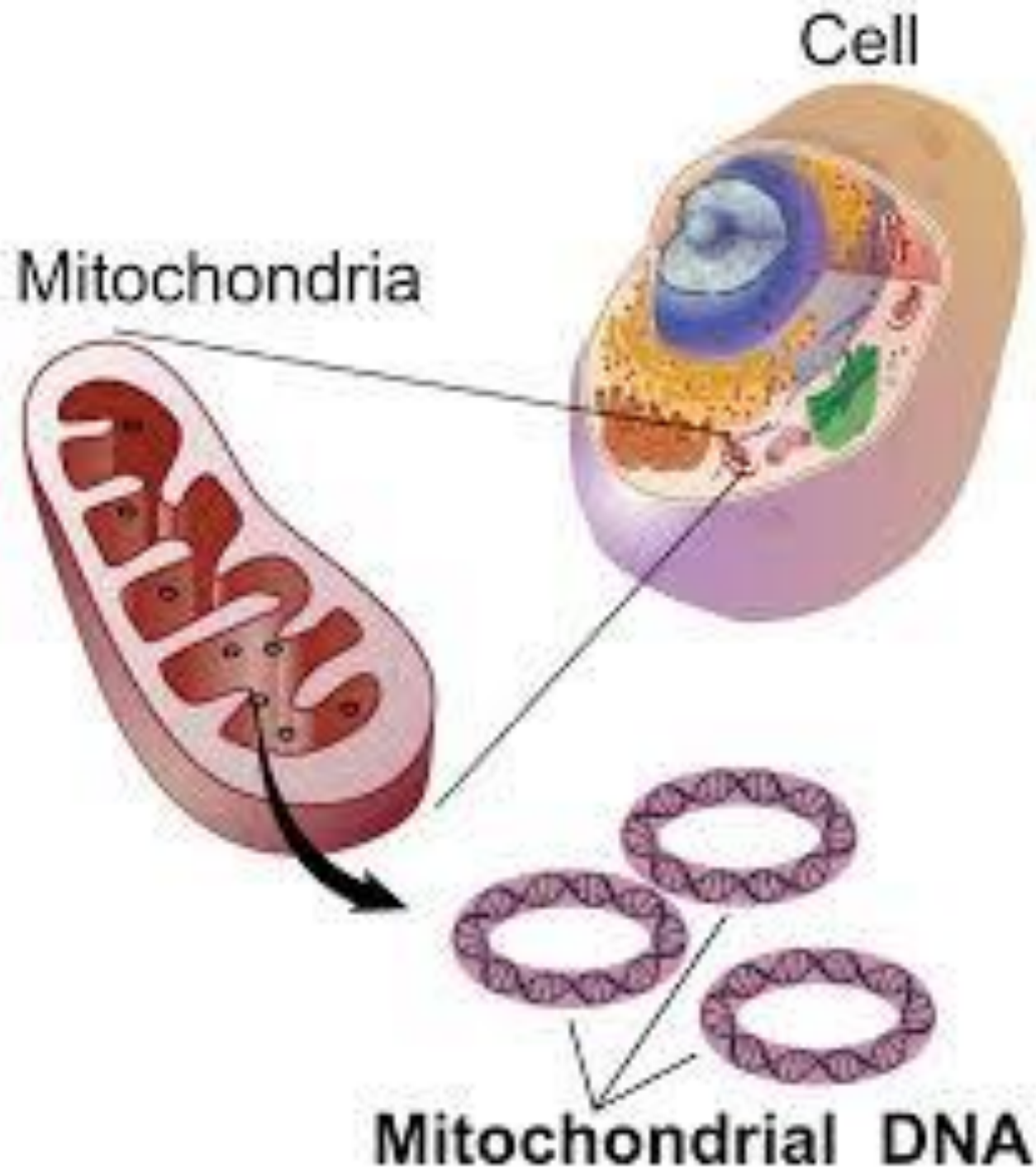
Consultant in Metabolic Medicine

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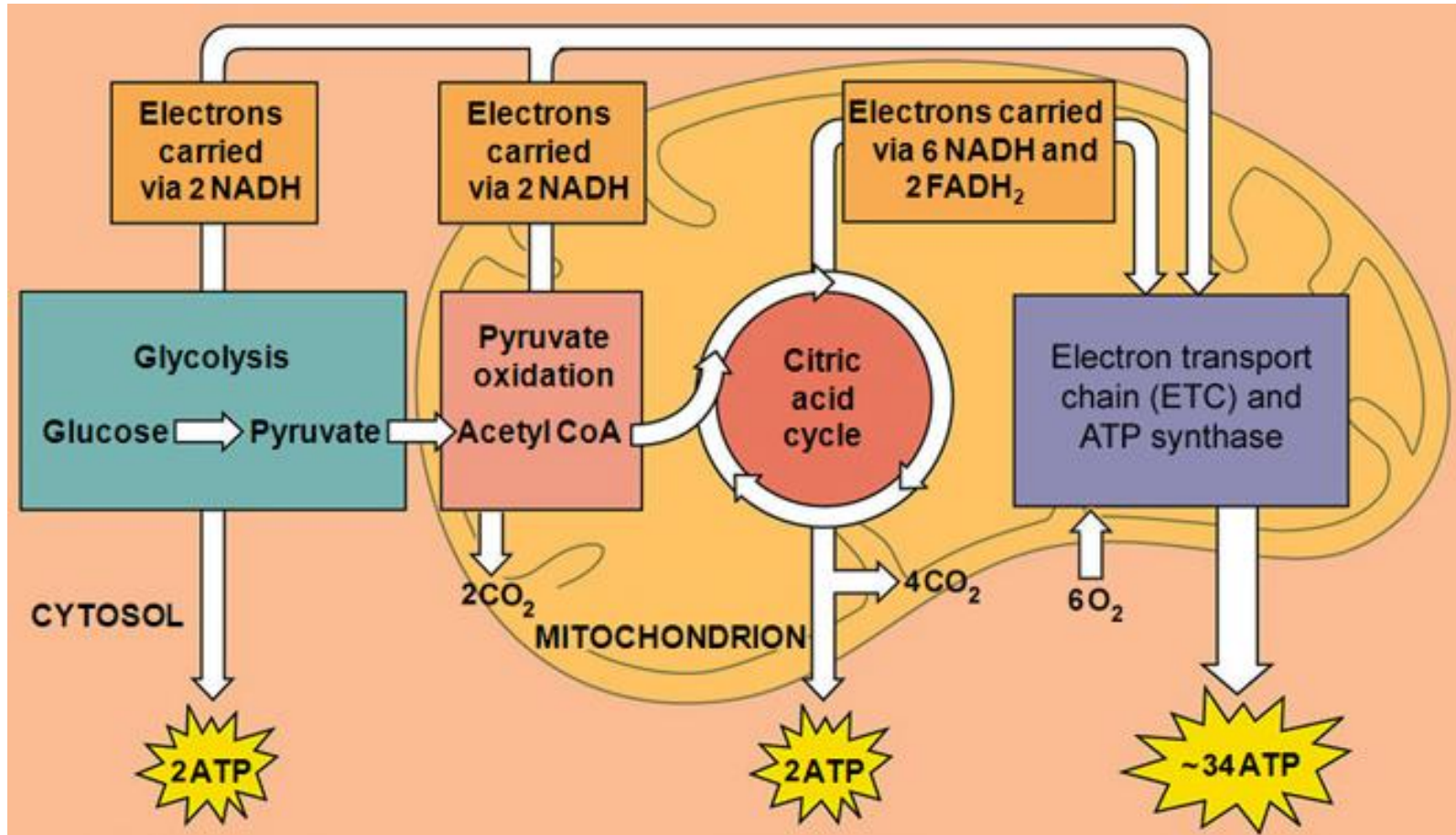
October 2018







# Mitochondria make energy in our cells











# Brain scans and nerve tests

- CT scans
- MRI scans
- Lumbar puncture
- Nerve conduction studies and EMG
- EEG



# Blood tests

- Lactic acid (lactate)
- CK (creatinine kinase)
- Diabetes tests
  - Glucose
  - HbA1c
- FGF-21 (fibroblast growth factor-21)

# Blood tests

- Lactic acid (lactate)
- CK (creatine kinase)
- Diabetes tests
  - Glucose
  - HbA1c
- FGF-21 (fibroblast growth factor-21)
- Kidney (renal) function tests
- Liver function tests
- Calcium, bone and vitamin D tests



# Cardiac tests

- ECG
- 24 hour Holter monitor (24 hour tape)
- Echocardiogram

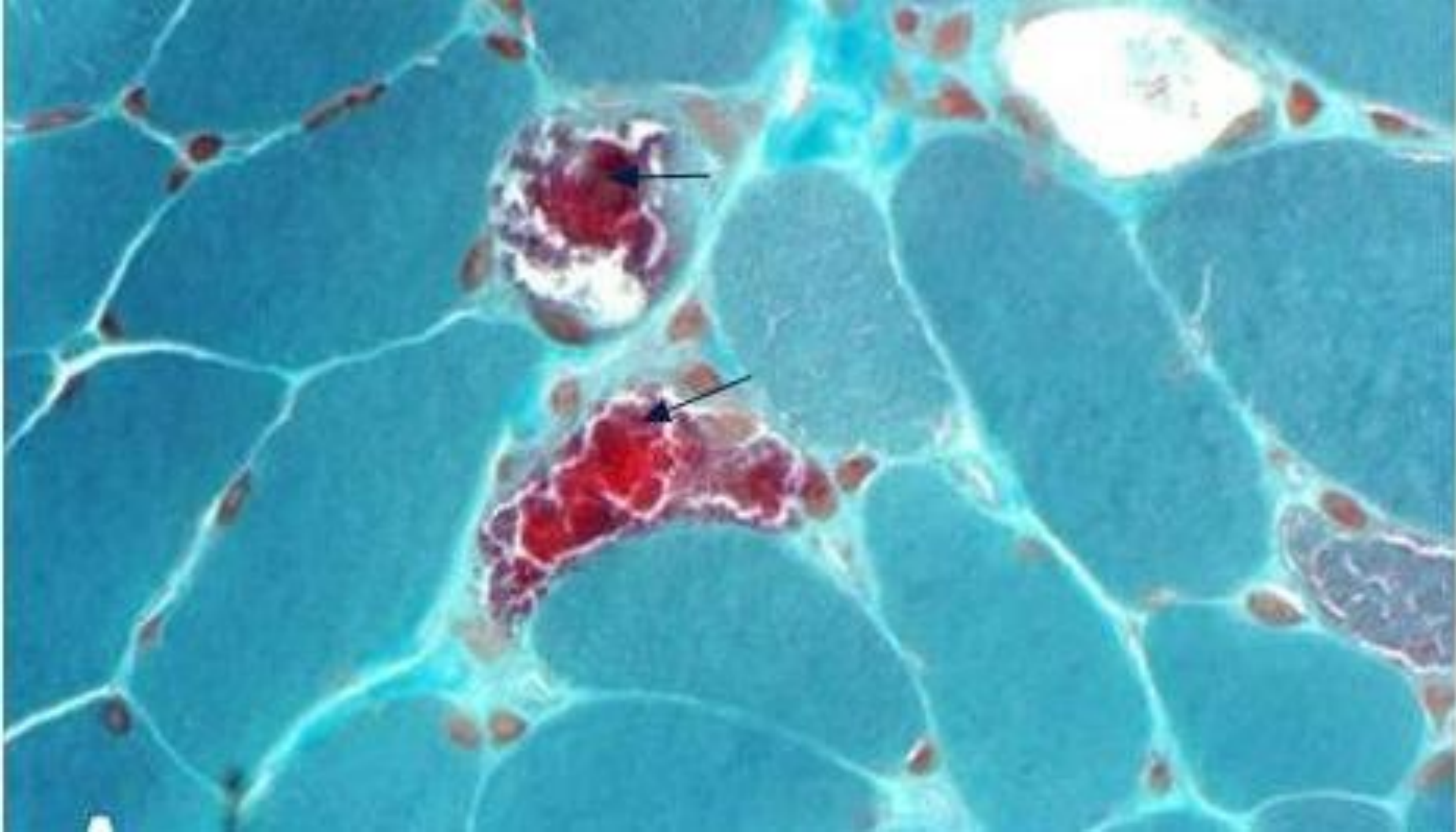
# Other tests

- Hearing tests (audiology)
- Eye movements, retina, optic nerve
- Swallowing tests, investigations of gut muscle



# Muscle biopsy

- Microscope
  - Ragged red fibres
  - COX negative fibres





# Muscle biopsy

- Microscope
  - Ragged red fibres
  - COX negative fibres
- Mitochondrial enzyme studies
- Mitochondrial genetic studies

# Genetic tests

- Samples
  - Blood
  - Urine
  - Saliva
  - Muscle
- Mitochondrial genetic mistake e.g.
  - m.3243, m.8344
- Nucleus genetic mistake e.g.
  - c.1399
- Human Genomes Project