

# Priorities for research in primary mitochondrial disease: patients' and clinicians' Top 10.

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## Background

Resources for research are limited. Consequently it is important to understand which are the most important questions for research to address from the point of view of patients, carers and healthcare professionals, and to target resources accordingly. The rarity of mitochondrial disease, and the diversity in how individuals are affected, are a further challenge to attracting research.

## Aim

This Priority Setting Partnership aimed to stimulate research by consulting with patients, carers and healthcare professionals about their most important unanswered questions relating to care, treatment, management and the natural history of mitochondrial disease, in children and adults. The project was advised by a James Lind Alliance (JLA) facilitator to ensure it followed the JLA process.

## Method

Steering group and protocol established

First survey: collected 709 questions from 147 people  
34% patients / 32% carers / 34% healthcare professionals

Analysis: group into themes, write single research questions  
Longlist of 42 questions

Evidence checking: none of the questions found to be already fully answered

Prioritisation survey: questions ranked by 166 people  
63% patients / 21% carers / 16% healthcare professionals

Workshop: 24 question shortlist debated, Top 10 agreed  
8 patients / 9 carers / 13 healthcare professionals from 9 disciplines

Report launched ahead of Rare Disease Day 2020

## Top 10

1. Could an understanding of the cellular and molecular processes in mitochondrial disease lead to **new treatments**?
2. Can the damage to cells caused by mitochondrial disease be **repaired** (e.g. to restore hearing, vision, or repair the pancreas)?
3. What are the biological mechanisms that cause mitochondrial disease to get **worse over time**?
4. What biomarkers (biological markers that can be measured e.g. in blood samples) could be used to **diagnose** mitochondrial disease and to track its progress?
5. Could **gene therapy** help people with mitochondrial disease?
6. What are the psychological impacts of mitochondrial disease? What are the best ways to provide **psychological support** for people with mitochondrial disease and their families?
7. What are the best ways to reduce the **risk of stroke-like episodes** in people with mitochondrial disease?
8. What factors could **trigger the start** of mitochondrial disease in people who have a genetic mutation?
9. Why are people with the same genetic mutation affected so **differently** in mitochondrial disease?
10. What are the most effective ways to **treat and manage fatigue**?

## Next steps

- Disseminate through project networks
- Liaise with research funders
- Publish as peer reviewed article



Priority Setting Partnerships



The PSP and its findings are endorsed by: EpiCARE, ERN-EYE, MetabERN and EURO-NMD.



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