

TOP 10 RESEARCH PRIORITIES FOR MITOCHONDRIAL DISEASE

- 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?

The research priorities were generated by patients, carers and healthcare professionals through an 18-month JLA Priority Setting Partnership, and we encourage:

- **research funders** to include these priorities in research strategies and to target these topics for future research funding;
- **researchers** to focus their efforts on the highest priority questions and to mention the JLA Mitochondrial PSP in their applications for funding.
- **funders, researchers and all interested parties** to share this Top 10 with others and to raise awareness of the need for more research on mitochondrial disease in the UK.

The PSP and its findings are endorsed by the following European Reference Networks: EpiCARE, ERN-EYE, Euro-NMD and MetabERN.



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