### The role of Nicotinamide Riboside in mitochondrial biogenesis

### Mitochondria are important parts of the cell that are responsible for producing energy. The amount of energy they produce depends on how much energy your body needs to function and this energy production can be severely impaired in people with mitochondrial disease. Symptoms of mitochondrial disease vary widely but usually involve the brain, nerves and muscles, as these are tissues that need a lot of energy. Mitochondrial disorders affect 1 in 5,000 of the UK population and there is currently no cure.

### Some scientists think that increasing the number of mitochondria in your body (mitochondrial biogenesis) might be an effective treatment for the symptoms of mitochondrial disease. Studies carried out in mice have shown that a type of B-vitamin called Nicotinamide Riboside (NR), is able to increase the number of mitochondria, leading to increased energy and a reduction in the symptoms of mitochondrial disease.

### The aim of this study is to investigate if the same B vitamin, Nicotinamide Riboside, can increase energy production and reduce symptoms in humans with mitochondrial disease.

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### The study will consist of two parts:

### Part 1: We will give participants a single oral dose of Nicotinamide Riboside and measure the levels of NR in their bloodstream at regular intervals. This will involve a single overnight stay and simple blood tests.

### Part 2: This requires 6 separate visits from each participant. We will carry out a series of standard tests including a muscle biopsy and an MRI scan, then ask participants to take a course of Nicotinamide Riboside (twice daily for 4 weeks). After 4 weeks of treatment we will carry out the same tests again to see if there have been any changes in response to the treatment.

**Patient profile:**

* **18-70 years, male or female**
* **Mitochondrial disease caused by a single mitochondrial DNA deletion, as well as a diagnosis of Progressive External Ophthalmoplegia (PEO) and exercise intolerance**

If you think you might be eligible and are interested in taking part in this research, or would like any further information, please contact the study team directly:

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