**Patient profile:**

**18-70 years, male or female**

* **Mitochondrial disease caused by mutations in mtDNA , e.g., MELAS or LHON**
* **Mitochondrial disease caused by mutations in *SPG7* or *POLG* genes**

**[11C]PK11195 PET as a biomarker to diagnose and monitor natural history in mitochondrial disease**

### 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 5000 of the UK population and there is currently no cure.

This is a small study to see if we can use the molecule [11C]PK11195 to diagnose and study the progression of mitochondrial disease. [11C]PK11195 is a radioactive compound which binds to certain proteins in the mitochondria. This interaction releases a signal that can be detected using a specialised positron emission tomography (PET) scanner. This allows us to estimate the amount of mitochondria in different regions of the brain and compare it to the brains of people who don’t have mitochondrial disease.

The study involves one screening visit (which we can carry out in the participant’s home) and one study visit at the Wolfson Brain Imaging Centre in Cambridge. We will inject a small amount of [11C]PK11195 into the participant’s bloodstream, then carry out a combined PET/MRI scan lasting roughly 90 minutes. After the scan, the study is complete and the participant is free to go home. The study visit shouldn’t last any longer than 3 hours in total.

The scan involves exposure to a small amount of ionising radiation. The radiation dose involved is similar to the average amount a person in the UK receives each year from background radiation sources, such as cosmic rays and sunlight, and is considered as low risk. [11C]PK11195 has been used in research for more than 15 years, without toxicity or other adverse reactions.

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:

|  |  |
| --- | --- |
| **Research Nurse: Miss Sarah Bird**Tel: 01223 331506Email: sc921@medschl.cam.ac.uk | **Study Coordinator: Miss Zoe McIntyre**Tel: 01223 331506Email: zm276@medschl.cam.ac.uk |