Lay Summary

BRAIN UK Ref: 19/014

PURA syndrome patient tissue sample banking for histopathological and molecular analysis

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PURA syndrome is a rare disorder in which development of the brain in young people causes them to have development problems, which include movement and learning ability. This disorder is caused by mutations, which is a change in the DNA sequence, in the Pur-alpha gene (PURA). Currently there are no treatments, except managing the disease, which causes difficulty in mobility, learning difficulty and epilepsy. Patients with the disease, require lifelong care, including in day to day activity. The way the mutation in the PURA gene occurs is not known currently.

In my lab we have been able to recreate an experimental mouse model that mimics the PURA syndrome and we have shown the mutation in the brain and other organs, similar to humans. In this study, we plan to use left over, and archived brain and other tissue from PURA syndrome and normal patients, to study the disease. We will look at levels of Pur-alpha and other associated genes, in normal and PURA syndrome tissue. This could lead to finding out, how the mutation occurs, and in the future, may lead to treatment, that could either reverse this or stop it all together.