Lay Summary

BRAIN UK Ref: 18/010

Identification of Differentially Expressed Proteins and Genes Impacting Seizures and Risk of SUDEP in Dravet Syndrome

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Dravet Syndrome is a severe form of epilepsy that is associated with an increased risk of sudden unexpected death in epilepsy (SUDEP). Although a gene mutation has been identified in 80% of patients, seizures generally remain drug resistant. Defects of various neurotransmitter signals in the brain have been implicated in Dravet Syndrome, however the processes that lead to seizures are still not well understood. We aim to evaluate these processes by identifying pathological differences in the proteins and genes of involved areas of the brain (hippocampus and brainstem). This will be done in Dravet Syndrome patients and compared to patients with and without epilepsy. We aim to improve our understanding of the processes associated with seizures in these brain regions. This will provide insight into potential therapeutic strategies for improving seizure management in Dravet Syndrome, reducing SUDEP risk and understanding of other forms of epilepsy.