Lay Summary

BRAIN UK Ref: 11/005

Fight Alpers'

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Alpers' syndrome is a rare progressive disease typically of childhood characterized by seizures, mental retardation, blindness and liver failure. It is caused by a defect in a gene present in subcellular elements called mitochondria leading to their dysfunction in the central nervous system and liver. Given the rapid onset of symptoms and a lack of cure diagnosis needs to be rapid in order to better manage the debilitating symptoms of this disease.

As Alpers' syndrome is rare there is limited knowledge of the neurodegenerative changes that occur. An increased understanding of the pathological processes will ultimately allow the development of better treatment strategies for affected patients. The applicant will use an array of special staining methods on tissue from Alper's syndrome patients in order to determine any link between brain pathology, mitochondrial dysfunction and clinical symptoms exhibited.

Publications:

Date	Publication title
2018	Dissecting the Neuronal Vulnerability Underpinning Alpers' Syndrome: a Clinical and Neuropathological Study