Lay Summary

BRAIN UK Ref: 19/004

Validation of histopathological findings in HTRA1 mutation carriers

Prof Martin Dichgans, Klinikum der Universität München, Munich, Germany

Stroke is the leading cause of long-term disability and the second most common cause of death. In about 20% of cases stroke is caused by changes in small brain vessels. Inherited defects in a gene called HTRA1 are a rare cause of stroke because of changes in brain vessels. We recently found in a mouse model that inherited defects in this gene cause deposition of specific proteins in brain vessels. To better understand what role these proteins, have in humans we plan to investigate brain specimens from patients who have inherited a defect in HTRA1 gene. From these studies we expect to obtain a better understanding how other types of stroke develop.