CARASIL – a window into Alzheimer’s disease?

https://neurodegenerationresearch.eu/survey/carasil-a-window-into-alzheimers-disease/

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Title of project or programme

CARASIL - a window into Alzheimer's disease?

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Alzheimer's Research UK

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€ 40,674

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Total duration of award in years

3

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Research Abstract
Cerebral autosomal recessive arteryopathy, with subcortical infarcts and leukoencephalopathy (CARASIL), is a rare, recessive disease characterized by early-onset changes in the white matter of the brain, associated with strokes and progressive dementia.

Eight mutations in the HTRA1 gene have been described as causes for CARASIL. Given that several cases do not harbor mutations in this gene, it is very likely that other genes also play a role in this disease.

To address this issue, we collaborated with Dr. Lohmann (Istanbul University and Tübingen University) and collected a cohort of 30 CARASIL-like Turkish cases. We will use recently developed sequencing technology to determine the sequence of the entire coding part of these

patients’ genome (the exome) and will determine the genetic causes of the disease.

We recently used this same approach to identify TREM2 mutations as the cause of frontotemporal dementia in three Turkish families and subsequently identified a heterozygous variant in the same gene as a risk factor for Alzheimer’s disease (AD).

This Pilot Project will allow the identification of novel genes causative of CARASIL. Given the overlap in pathways between CARASIL and AD, these genes will be excellent targets for future genetic analyses of risk factors in AD.

Further information available at:

Types:
Investments < €500k

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United Kingdom

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