The genetics of frontotemporal dementia

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(Medical Xpress)—Professor Stuart Pickering-Brown, a world expert in the disease from the University of Manchester, will shed new light on a gene that causes the disease on Tuesday (25 March) at Alzheimer's Research UK Conference 2014 in Oxford. The study has revealed new information about some of the features of the disease.

Alzheimer's Research UK is the UK's leading dementia research charity, funding more than £20m of pioneering research into the condition across the UK. The charity's annual conference, which takes place on 25 and 26 March, is the largest of its kind in the UK and will see leading scientists share their progress in the drive to defeat dementia.

Prof Pickering-Brown and his team are investigating a gene called C9orf72, which has been implicated in the development of frontotemporal dementia (FTD). This relatively rare form of dementia, which usually affects people under 65, causes distressing symptoms including personality and behavioural changes, loss of ability to reason, and problems with language.

Up to 40% of people with FTD have an inherited form of the disease, and it's thought around 9% of cases in the UK are caused by a faulty version of the C9orf72 gene. Earlier research has shown that this gene produces repeated protein fragments called 'dipeptides', and the team in Manchester is investigating whether these are involved in causing the disease. Using state-of-the-art techniques, the researchers have been able to reproduce these protein fragments in cells in order to study them in the lab. Their research has shown that the fragments accumulate inside

cells, and the team is now working to understand whether they are harmful to cells.

Professor Pickering-Brown said: "To be able to develop new treatments for people with FTD, it's important to understand the biological mechanisms involved in the disease. The faulty gene we are studying has a number of different biological effects, and we want to understand which of these play a role in the disease, and how. By recreating some of the effects we see in the brain, our team has been able to produce a valuable tool to help us solve this difficult puzzle. Importantly, our research could also enable us to test the effects of potential new treatments for the disease in the future."

Dr Simon Ridley, Head of Research at Alzheimer's Research UK, said: "When this faulty gene was revealed as one cause of FTD, the discovery raised a number of important questions about how the gene causes damage, and how this damage could be stopped. This useful study has begun to answer some of those questions and opened new avenues for research. It will now be essential to follow up this research to understand how these protein fragments may be involved in FTD, and whether treatments designed to target them could have a beneficial effect. For results like these to be translated into benefits for people, it's crucial that we continue investing in research."

Provided by University of Manchester