# Scientists develop refined diagnostic tool for inherited dementias

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Researchers at the Medical Research Council (MRC) have developed and tested a more efficient and cost effective way to diagnose early onset, genetic forms of dementia.

Scientists and doctors at the MRC Prion Unit and the National Institute of Health Research (NIHR) Queen Square Dementia Biomedical Research Unit (BRU) have developed an integrated diagnostic tool (the 'MRC Dementia Gene Panel') which brings together for the first time all 17 genes known to play a substantial role in causing inherited forms of dementia.

Using a modern method of analysing the genetic material or DNA from a patient's blood sample, called 'next generation sequencing', they were able to look for abnormalities in all of these genes simultaneously, quickly and cheaply. Their work, published online this week in the journal *Neurobiology of Aging*, shows that the technology is highly accurate and comprehensively identified the genetic abnormalities.

Dementia is one of the most important healthcare challenges worldwide. Some forms of dementia, particularly in younger people, run in families and are caused by one or more faulty genes being passed down from parent to child. While these forms are relatively uncommon and are currently thought to account for around two per cent of all dementias, they can be detected and diagnosed definitively by genetic testing. The technique will be used at University College London Hospitals NHS Foundation Trust (UCLH) to offer improved diagnosis to patients in

their neurogenetics clinics. The researchers will also use this technology to study further the relevance of these genes as a tool in the diagnosis of patients with dementia, particularly for those developing dementia before the age of 65 where a genetic cause of their illness is more likely.

There has been significant progress in the understanding of the genetic causes of dementia in recent years. Genetic mutations in more than 17 genes are known to cause specific types of dementias while other changes in these and other genes can increase a person's risk of developing dementia over the course of their lifetime. Although these genes account for only a small minority of the total number of patients with dementia, a simple blood test could provide a definitive diagnosis, and may lead to the opportunity for blood relatives to find out if they share an increased risk of developing the disease.

Dr Simon Mead, who led the research at the MRC Prion Unit and the NIHR Dementia BRU, said,

"We have developed this technology because we know many patients don't get an accurate diagnosis. We hope that this and related technologies will greatly improve the way we identify patients with genetic forms of dementia. Currently the tests for dementia genes are not widely available for doctors and, where they are available, the tests have been very expensive and it is hard sometimes to know who needs to be tested. What is really needed is a simple and cost effective way to screen all genes at the same time, which could be used in a larger proportion of patients. It is likely that screening more patients in this way will pick up genetic causes that might otherwise be missed."

Professor Martin Rossor, BRU Director, said,

"'This technology will revolutionise our ability to provide a rapid and cost effective diagnosis. It will be particularly important for those

patients with a diagnosis of young onset dementia and their families. There is estimated to be over 18,000 people in the UK below the age of 65 with dementia and it is this young onset group that often has a family history of disease."

Dr Catherine Elliott, Head of Neurosciences and Mental Health at the MRC, said:

"Tackling dementia is a priority for the MRC and improving methods of diagnosis is crucial for truly understanding the burden of these devastating diseases. This is a significant development in applying new technology to improve diagnosis in those groups of patients with genetic causes of dementia."

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