FACTSHEET 2

WHAT IS FAMILIAL FRONTOTEMPORAL DEMENTIA?

Frontotemporal dementia or FTD is a progressive disorder of the brain. It can affect behaviour, language skills and movement.

For many people with FTD, the cause is not known. In around a third of people however it can be triggered by a genetic problem – we call this familial FTD.

The main genes involved are called:

- Tau or MAPT
- Progranulin or GRN
- C9orf72

There are also some other rare genetic causes of FTD which are uncommon. These genes are called:

- TBK1
- TARDBP
- VCP
- CHMP2B
- FUS
- SQSTM1

Some families have a clear family history of FTD but no abnormal genes have been identified at present.

How likely am I to inherit FTD?

In people with abnormalities in these genes there is a 50 per cent chance that the abnormal version of the gene will be passed on to their children.However there are a couple of important things to know:

- The age at which symptoms of FTD begin is very variable. Some people develop FTD around the same age that their parent developed it. However for other people there can up to twenty years difference in the age that FTD develops.
- In some of the genes, particularly GRN and C9orf72, there can be something that is called age-related penetrance. This means that although someone may carry the genetic mutation they may only develop FTD in older age or not at all.

How is familial FTD diagnosed?

Usually a diagnosis of FTD has already been made by a specialist by performing a series of tests including a scan of the brain.

A genetic test can then be performed on a blood sample in people who the specialist thinks may have a genetic cause of their FTD.

The test is usually done because a number of other family members have FTD or an associated disorder. However it is also done by some specialists when symptoms or brain scans suggest familial FTD.

What can I do if a relative has familial FTD?

In families who are known to have a gene abnormality, expert genetic advice is important.

If you are worried about your risk of developing familial FTD or wish to discuss it further, your GP or primary care physician will be able to refer you to a specialist genetics service to discuss your concerns and arrange genetic testing if needed

The results of any genetic test have important implications not only for the person being tested but also for the rest of the family. It is important to discuss these carefully with someone who has expertise in genetic counselling, and also with the family, before any genetic tests are performed.

What support is available?

The Familial FTD support group has been set up to help members of families with familial FTD.

The aim is to provide the opportunity for people to find out more information about familial FTD and to meet other similar families.

The website can be found at www.raredementiasupport.org where more information is available.

Another useful organization is the Genetic Alliance – www.geneticalliance.org.uk

Is there research into familial FTD?

The Genetic FTD Initiative or GENFI is a group of research centres across Europe and Canada studying familial FTD.

The aim is to gain a better understanding of the disorder using brain imaging, blood and spinal fluid tests, and psychology testing. GENFI studies people who are over the age of 18 and have a first-degree relative with familial FTD. More information is available at www.genfi.org

A similar study in the US is called ALLFTD, and in Australia is called DINAD – more information is available on the website of the FTD Prevention Initiative or FPI, which brings together all of the familial FTD studies in the world: www.thefpi.org