FACTSHEET 1

WHAT IS FRONTOTEMPORAL DEMENTIA?

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

The brain is made up of four lobes – the frontal lobe, parietal lobe, temporal lobe and occipital lobe. In FTD, there is progressive loss of cells in the frontal and temporal lobes of the brain.

The main subtypes of FTD are called:

- Behavioural variant FTD or bvFTD
- Primary progressive aphasia or PPA

PPA can be further split into three main subtypes known as:

- semantic variant, also known as semantic dementia or SD
- nonfluent variant, also known as progressive nonfluent aphasia or PNFA
- logopenic variant, also known as logopenic aphasia or LPA

What causes FTD?

For most people, the cause of FTD is not known

FTD can affect both men and women. It usually affects people in their 50s or 60s, although it can affect people both younger and older than this.

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

See FACTSHEET 2 for more details on familial FTD.

What are the symptoms of FTD?

Damage to the frontal and temporal lobes causes problems with behaviour and language skills.

Behavioural variant FTD

In bvFTD the initial symptoms are usually a change in personality or behaviour. These may include:

- Behaving inappropriately
- Becoming less interested in things or in people
- Becoming more obsessive or repetitive
- Changes in the types of food eaten
- Problems with planning or problem solving
- Difficulty concentrating

For more details about bvFTD see FACTSHEET 3.

Primary progressive aphasia

In PPA the initial symptoms are problems with language skills. For more details about PPA see FACTSHEET 4.

Semantic variant

In the early stages there may be subtle changes in the ability to find or understand words. For more details see FACTSHEET 5.

Nonfluent variant

The main problem is usually producing speech. Speech may become slow and hesitant, and words may be missed out or pronounced incorrectly. For more details see FACTSHEET 6.

Logopenic variant

People may pause in the middle of sentences and have difficulties finding words.

Do people with FTD have physical problems?

It is uncommon to have physical problems in the early stages but some people may also develop problems with movement:

- Motor neurone disease – see FACTSHEET 7
- Parkinsonism where there are symptoms similar to Parkinson's disease – see FACTSHEET 8. The two main types of parkinsonism seen are called corticobasal syndrome or CBS, and progressive supranuclear palsy or PSP. See FACTSHEETS 9 and 10 for more details.

How is FTD diagnosed?

Usually a diagnosis is made by a specialist rather than a GP. There is no single test that will make a diagnosis except in people who have a genetic cause. A series of tests are usually performed including a scan of the brain. Diagnosis is discussed in more details in FACTSHEET 11.

Is there a treatment for FTD?

There is currently no cure for FTD but there are some important things which can help when caring for someone. These include:

- Strategies for dealing with behavioural symptoms
- Speech and language therapy for people with PPA
- Genetic advice and counselling for people with familial FTD
- These are discussed in more detail in FACTSHEET 12.

What is the prognosis of FTD?

The rate of progression is very variable and can range from two to twenty years. As the disease progresses, people will need help with most aspects of daily life. People may need nursing care.

What support is available?

Useful organizations that can provide information include:

- Rare Dementia Support (UK)
  www.raredementiasupport.org
- Alzheimer's Society (UK)
  www.alzheimers.org.uk
- Association for Frontotemporal Degeneration (US)
  www.theaftd.org
- The Australian FTD Association (Australia)
  www.theaftd.org.au
- For links to other support groups around the world – www.worldftdunited.net

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

FACTSHEET 3

WHAT IS BEHAVIOURAL VARIANT FTD?

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

Behavioural variant FTD is one of the subtypes of FTD. It is also known as bvFTD. The first symptoms are usually changes in behaviour or a change in personality. In the early stages these changes may be very subtle and not be noticed as abnormal at first.

Symptoms can include:

Behaving inappropriately – disinhibition

- Losing normal inhibitions
- Losing manners
- Being more extrovert than previously
- Making inappropriate comments to people
- Acting more impulsively such as spending excessive amounts of money
- More or inappropriate interest in sex

Becoming less interested in things – apathy

- Less motivation to do things
- Being more passive than before
- Needing prompting to do routine activities
- Paying less attention to personal hygiene and dress
- Loss of interest in sex

Becoming less interested in people – loss of empathy

- Being less sympathetic to others than before
- Interacting less with people
- Withdrawing from social activities

Becoming more obsessive or repetitive in their behaviour

- Being fixed in the way they do things
- Having to stick to routines
- Saying things repetitively
- Collecting or hoarding things

Changes in the types of food eaten, such as sweet foods

- Increased craving for certain foods, often sweet things
- Food fads – eating one specific type of food
- Binge eating
- Increased consumption of alcohol or cigarettes
- Eating very quickly

Usually the person will be unaware of the true extent of their problems and lack insight into what is happening to them.

As well as changes in behaviour, there may be problems with thinking as well:

- In the early stages of the disease this may be problems in planning, organisation, making decisions or solving problems.
- People may have difficulty concentrating on one thing and seem very distractible

As the disease progresses problems with behaviour will tend to become worse. However some behaviours such as disinhibition may become less of a problem.

Other parts of thinking may also become affected over time such as:

- Finding the right word
- Understanding speech
- Remembering things

FACTSHEET 3
WHAT IS BEHAVIOURAL VARIANT FTD?

Does bvFTD run in families?

In about a third of cases bvFTD can be a genetic disorder and run in families. Mutations in the tau, progranulin or C9orf72 genes are the most common. See FACTSHEET 2 for more details about familial FTD.

How is bvFTD diagnosed?

Usually a diagnosis is made by a specialist rather than a GP. There is no single test that will make a diagnosis except in some people who have a genetic cause. A series of tests are usually performed including a scan of the brain – see FACTSHEET 11 for more details.

Is there a treatment for bvFTD?

There is currently no cure for bvFTD but there are some important things which can help when caring for someone – see FACTSHEET 12 for more details.

What is the prognosis of bvFTD?

The rate of progression is very variable and can range from two to twenty years. As the disease progresses, people will need help with most aspects of daily life. Continence may become a problem and people commonly develop problems with swallowing. Eventually nursing care may become necessary.

What support is available?

Useful organizations that can provide information include:

- Rare Dementia Support (UK)
  www.raredementiasupport.org
- Alzheimer's Society (UK)
  www.alzheimers.org.uk
- Association for Frontotemporal Degeneration (US)
  www.theaftd.org
- The Australian FTD Association (Australia)
  www.theaftd.org.au
- For links to other support groups around the world – www.worldftdunited.net

FACTSHEET 4

WHAT IS PRIMARY PROGRESSIVE APHASIA?

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

Primary progressive aphasia or PPA is one of the subtypes of FTD. The three main subtypes of PPA are called:

- semantic variant or svPPA
- nonfluent variant or nfvPPA
- logopenic variant or lvPPA

These are also sometimes called:

- Semantic dementia or SD
- Progressive nonfluent aphasia or PNFA
- Logopenic aphasia or LPA

In all of the types of PPA impairment of language skills is the main problem initially.

Up to 20% of people do not fit into one of the three main subtypes, and are often just called PPA, or sometimes mixed PPA, or PPA-not otherwise specified.

Symptoms of svPPA include:

- Difficulty finding the right word
- Losing understanding of what words mean
- Talking about things in a vague manner
- Difficulty understanding what other people are saying
- Problems with reading
- Problems with spelling

See FACTSHEET 5 for more details.

Symptoms of nfvPPA include:

- Slow, hesitant speech
- Difficulty finding the right word to say
- Pronouncing words incorrectly
- 'Telegraphic' speech
- Producing the wrong grammar
- Saying the opposite word to the one they mean to say
- Problems with reading
- Problems with spelling

See FACTSHEET 6 for more details.

Symptoms of lvPPA include:

- Difficulty finding the right word to say
- Pausing in the middle of sentences
- Difficulty repeating sentences

LvPPA is usually felt to be an unusual form of Alzheimer's disease, and is sometimes called the language variant of Alzheimer's disease.

In all of the different forms of PPA other non-language symptoms occur later on, including:

- Changes in behavior
- Problems with planning and problem solving
- Difficulty remembering things

Physical problems occur in some people similar to the symptoms of Parkinson's disease. Very rarely people with PPA may develop motor neurone disease.

Does PPA run in families?

In some cases PPA can be a genetic disorder and run in families. This is most likely for nfvPPA. Mutations in either the progranulin or C9orf72

genes can cause PPA.

See FACTSHEET 2 for more details about familial FTD.

How is PPA diagnosed?

Usually a diagnosis is made by a specialist rather than a GP. There is no single test that will make a diagnosis except in some people who have a genetic cause. A series of tests are usually performed including a scan of the brain.

See FACTSHEET 11 for more details.

Is there a treatment for PPA?

There is currently no cure for PPA but there are some important things which can help when caring for someone – see FACTSHEET 12 for more details.

The PPA Support Group has been set up to help people with PPA and their carers. It meets a number of times a year. More information can be found at www.raredementiasupport.org

FACTSHEET 5

WHAT IS SEMANTIC VARIANT PPA?

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

The main subtypes of FTD are called behavioural variant FTD and primary progressive aphasia or PPA. PPA can be further split into three main subtypes, one of which is called semantic variant PPA or semantic dementia.

The first symptoms of svPPA are usually very subtle changes in finding words or understanding them. This is thought to be due to loss of semantic knowledge, our understanding of what things are or their meaning.

Symptoms can include:

Difficulty finding the right word

People may use another word instead of the correct one, for example saying 'cat' instead of 'dog', or calling something a 'thing' or other vague term.

Losing understanding of what words mean

People may ask the meaning of a word that they have previously known.

Talking about things in a vague manner

People may not seem to be a making any sense when they speak.

Difficulty understanding what other people are saying

Sometimes this can be put down to people seeming to be 'deaf'.

Problems with reading

Problems with spelling

People may initially complain that they have a memory problem or that they have forgotten words. However unlike in Alzheimer's disease, day-to-day memory for events or the ability to find their way around are not usually affected in the early stages.

As the disease progresses other thinking problems may occur such as:

- Inability to recognise objects
- Inability to recognise faces
- Impairment of the memory for day-to-day events

Behavioural problems may also occur similar to those seen in behavioural variant FTD. In particular, people may:

- Behave inappropriately
- Become more obsessive or repetitive in their behaviour
- Develop a craving for sweet foods

See FACTSHEET 3 for more details on behavioural variant FTD.

Does svPPA run in families?

It is rare for svPPA to run in families. It usually occurs as a sporadic disorder without any genetic cause.

See FACTSHEET 2 for more details about familial FTD.

How is svPPA diagnosed?

Usually a diagnosis is made by a specialist rather than a GP. There is no single test that will make a diagnosis. A series of tests are usually performed including a scan of the brain. In svPPA the scan

may show loss of cells in the middle of the brain in an area called the temporal lobe.

See FACTSHEET 11 for more details.

Is there a treatment for svPPA?

There is currently no cure for svPPA but there are some important things which can help when caring for someone – see FACTSHEET 12 for more details.

The PPA Support Group has been set up to help people with PPA and their carers. It meets a number of times a year. More information can be found at www.raredementiasupport.org

FACTSHEET 6

WHAT IS NONFLUENT VARIANT PPA?

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

The main subtypes of FTD are called behavioural variant FTD and primary progressive aphasia or PPA. PPA can be further split into three main subtypes, one of which is called nonfluent variant PPA or progressive nonfluent aphasia.

The first symptoms of nfvPPA are usually a subtle change in the ability to produce speech. At first this might be only in certain circumstances such as when speaking on the telephone or when speaking in public.

Symptoms can include:

Slow, hesitant speech

Speech may seem effortful to produce and people may seem to stutter before they can get the right word out. This is known as apraxia of speech.

Difficulty finding the right word to say

Pronouncing words incorrectly

People may say the wrong word, often similar in sound to the one they meant to say, for example 'aminal' instead of 'animal'.

'Telegraphic' speech

People may miss out the small words in sentences such as 'the' or 'and'.

Producing the wrong grammar

People may use the wrong tense such as saying 'happened' when they mean 'happen' – this is known as agrammatism.

Saying the opposite word to the one they mean to say

For example saying 'yes' when they mean 'no'.

Problems with reading

Problems with spelling

Understanding of speech is usually not affected in the early stages.

Non-language functions such as memory also tend not to be affected early on. However, as the disease progresses other thinking problems may occur such as:

- Difficulty understanding speech
- Problems with planning and problem solving

Behavioural problems similar to those seen in behavioural variant FTD may also occur although these often do not occur until the later stages of the disease.

Physical problems occur in some people similar to the symptoms of Parkinson's disease. Very rarely people with PPA may develop motor neurone disease.

It can be a frustrating condition and people may become depressed because of their inability to speak.

Does nfvPPA run in families?

In some cases nfvPPA can be a genetic disorder and run in families. Mutations in the progranulin, C9orf72 or TBK1 genes can cause nfvPPA.

See FACTSHEET 2 for more details about familial FTD.

How is nfvPPA diagnosed?

Usually a diagnosis is made by a specialist rather than a GP. There is no single test that will make a diagnosis except in some people who have a genetic cause. A series of tests are usually performed including a scan of the brain. In nfvPPA the scan may show loss of cells in the front of the brain in an area called the frontal lobe.

See FACTSHEET 11 for more details.

Is there a treatment for nfvPPA?

There is currently no cure for nfvPPA but there are some important things which can help when caring for someone – see FACTSHEET 12 for more details.

The PPA Support Group has been set up to help people with PPA and their carers. It meets a number of times a year. More information can be found at www.raredementiasupport.org

FACTSHEET 7

WHAT IS FTD WITH MOTOR NEURONE DISEASE?

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

Motor neurone disease or MND is a disorder of the nerves that control the body's motor function. It affects the nerves that go to the muscles in the arms and legs, and that allow people to speak and swallow.

It is also known as amyotrophic lateral sclerosis or ALS.

About 1 in 10 people with FTD will also develop motor neurone disease. This is known as FTD-MND or FTD-ALS.

It occurs usually in people who have behavioural variant FTD and is less common in people with nfvPPA. It is very rare in people with svPPA.

It occurs more commonly in people who have a mutation in the C9orf72 gene.

MND may present either before or after the symptoms of FTD. In other words, some people who have MND as their first symptom may go on to develop FTD. Similarly, some people who have FTD as their first symptom may go on to develop MND.

In people who have FTD-MND, symptoms may progress more rapidly than in those people with FTD alone, although this is not the case for every person.

Symptoms vary from person to person, but can include:

- Wasting and weakness of muscles
- Twitching of muscles (called fasciculations)
- Stiffness of muscles
- Problems with articulation (production of speech) such that the speech may sound slurred
- Problems with swallowing

Later on in the disease some people develop problems with their breathing.

Does FTD-MND run in families?

FTD-MND can sometimes run in families. Some members of the family may have MND without FTD, or vice versa. This is usually due to a problem in the C9orf72 gene.

See FACTSHEET 2 for more details about familial FTD.

How is FTD-MND diagnosed?

Usually a diagnosis is made by a specialist rather than a GP.

For FTD, there is no single test that will make a diagnosis except in some people who have a genetic cause. A series of tests are usually performed including a scan of the brain.

A diagnosis of MND may be made from the symptoms and signs found on clinical examination. However the diagnosis is usually supported by a series of tests including a needle test of the muscle electrical activity called electromyography or EMG.

See FACTSHEET 11 for more details.

Is there a treatment for FTD-MND?

There is currently no cure for FTD-MND but there are some important things which can help when caring for someone – see FACTSHEET 12 for more details.

People with MND are commonly given a drug called riluzole which has been shown to have some mild benefit. This will usually be started by a specialist in motor neurone disease.

Useful organizations that can provide information about FTD include:

- Rare Dementia Support (UK)
  www.raredementiasupport.org
- Alzheimer's Society (UK)
  www.alzheimers.org.uk
- Association for Frontotemporal Degeneration (US)
  www.theaftd.org
- The Australian FTD Association (Australia)
  www.theaftd.org.au
- For links to other support groups around the world – www.worldftdunited.net

For motor neurone disease, the Motor Neurone Disease Association can provide information and support to people diagnosed with MND – www.mndassociation.org

FACTSHEET 8

WHAT IS FTD WITH PARKINSONISM?

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

Parkinsonism is a disorder that affects movement. People may have greater difficulty moving or be slower in their movements – this is called bradykinesia. There may be stiffness in the arms and legs – this is called rigidity. People may also have too much movement such as a tremor where the hands shake.

Parkinson's disease is the most common form of parkinsonism. Some patients with FTD develop very similar symptoms. This is usually called FTD with parkinsonism or FTDP.

It occurs usually in people who have behavioural variant FTD or nfvPPA. It is very rare in people with svPPA.

There are two diseases in particular which may overlap with FTD and are often known as atypical parkinsonism. These are called:

- corticobasal syndrome or CBS, also known as corticobasal degeneration or CBD
- progressive supranuclear palsy or PSP

Corticobasal syndrome

Symptoms usually start on one side of the body but may go on to affect both sides.

They include the following symptoms that may affect the arm, leg or both:

- Slowness of movements
- Stiffness
- Tremor
- Jerkiness – this is called myoclonus
- Holding the arm in an odd posture – this is called an 'alien limb'
- Inability to do complex actions with the hands – this is called a limb apraxia

CBS may present before or after the onset of symptoms of frontotemporal dementia.

See FACTSHEET 9 for more details.

Progressive supranuclear palsy

Symptoms of PSP include:

- Difficulty walking
- Balance problems
- Recurrent falls that are often backwards
- Stiffness of the muscles – particularly the neck and trunk muscles
- Difficulty moving the eyes up and down – this may not be noticed by the person themselves or their family but by the doctor when they are examined

PSP may present before or after the onset of symptoms of frontotemporal dementia.

See FACTSHEET 10 for more details.

Does FTDP run in families?

FTDP can sometimes run in families. Some members of the family may have parkinsonism without FTD, or vice versa.

This can be due to a problem in one of the genes that causes familial FTD such as tau, progranulin or C9ORF72.

See FACTSHEET 2 for more details about familial FTD.

How is FTDP diagnosed?

Usually a diagnosis is made by a specialist rather than a GP. See FACTSHEET 11 for more details.

For FTD, there is no single test that will make a diagnosis except in some people who have a genetic cause. A series of tests are usually performed including a scan of the brain.

A diagnosis of parkinsonism may be made from the symptoms and signs found on clinical examination. However some tests may also be performed including a brain scan.

Is there a treatment for FTDP?

There is currently no cure for FTDP but there are some important things which can help when caring for someone – see FACTSHEET 12 for more details.

Drugs used in Parkinson's disease such as levodopa or other drugs that increase dopamine levels tend to have only a small or no effect on the symptoms of parkinsonism.

Useful organizations that can provide information about FTD include:

- Rare Dementia Support (UK)
  www.raredementiasupport.org
- Alzheimer's Society (UK)
  www.alzheimers.org.uk
- Association for Frontotemporal Degeneration (US)
  www.theaftd.org
- The Australian FTD Association (Australia)
  www.theaftd.org.au
- For links to other support groups around the world – www.worldftdunited.net

For parkinsonism the following organizations can provide information and support:

- PSP Association – www.pspassociation.org.uk

FACTSHEET 9

WHAT IS CORTICOBASAL SYNDROME?

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

FTD commonly overlaps with parkinsonian disorders causing problems with movement. One of these is called corticobasal syndrome or CBS. It is also sometimes known as corticobasal degeneration or CBD.

In CBS the first symptoms are often those that affect movement. However in some cases these can be preceded by behavioural or language symptoms, and some people may initially carry a diagnosis of behavioural variant FTD or PNFA.

Symptoms affecting movement usually start on one side of the body but may go on to affect both sides. They include the following symptoms that may affect the arm, leg or both:

- Stiffness – this is called rigidity
- Slowness of movements – this is called bradykinesia or akinesia
- Jerkiness – this is called myoclonus
- Holding the arm in an odd posture – this is called dystonia
- Inability to do complex actions with the hands – this is called a limb apraxia
- the limb seeming to move on its own – this is called an alien limb phenomena

Does CBS run in families?

In some cases CBS can be a genetic disorder and run in families. Mutations in either the progranulin or tau gene can cause CBS.

See FACTSHEET 2 for more details about familial FTD.

How is CBS diagnosed?

Usually a diagnosis is made by a specialist rather than a GP. See FACTSHEET 11 for more details.

A diagnosis of CBS may be made from the symptoms and signs found on clinical examination. However some tests may also be performed including a brain scan.

Is there a treatment for CBS?

There is currently no cure for CBS but there are some important things which can help when caring for someone – see FACTSHEET 12 for more details.

Drugs used in Parkinson's disease such as levodopa or other drugs that increase dopamine levels tend to have only a small or no effect on the movement symptoms.

The PSP Association has more information on CBS at www.pspassociation.org.uk

FACTSHEET 10

WHAT IS PROGRESSIVE SUPRANUCLEAR PALSY?

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

FTD commonly overlaps with parkinsonian disorders causing problems with movement. One of these is called progressive supranuclear palsy or PSP.

In PSP the first symptoms are often those that affect movement. However in some cases these can be preceded by behavioural or language symptoms, and some people may initially carry a diagnosis of behavioural variant FTD or PNFA.

Symptoms include:

- Balance problems
- Recurrent falls that are often backwards
- Difficulty walking
- Stiffness of the muscles – particularly the neck and trunk muscles
- Slowness of movements
- Difficulty moving the eyes up and down – this may not be noticed by the person themselves or their family but by the doctor when they are examined

Later on symptoms include:

- Slurring of speech
- Difficulty swallowing
- Laughing or crying at inappropriate times – this is called emotional lability

Does PSP run in families?

It is very unusual for PSP to be a genetic disorder and run in families. On rare occasions mutations in the tau gene can cause PSP.

See FACTSHEET 2 for more details about familial FTD.

How is PSP diagnosed?

Usually a diagnosis is made by a specialist rather than a GP. See FACTSHEET 11 for more details.

A diagnosis of PSP may be made from the symptoms and signs found on clinical examination. However some tests may also be performed. For example, specific changes can sometimes be seen on a brain scan called an MRI that can help support a diagnosis of PSP.

Is there a treatment for PSP?

There is currently no cure for PSP but there are some important things which can help when caring for someone – see FACTSHEET 12 for more details.

Drugs used in Parkinson's disease such as levodopa or other drugs that increase dopamine levels tend to have only a small or no effect on the movement symptoms.

The PSP Association has more information on PSP at www.pspassociation.org.uk

FACTSHEET 11

HOW IS FTD DIAGNOSED?

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

The main subtypes of FTD are called:

- Behavioural variant FTD or bvFTD
- Primary progressive aphasia or PPA

PPA can be further split into three main subtypes known as:

- semantic variant or semantic dementia
- nonfluent variant or progressive nonfluent aphasia
- logopenic variant or logopenic aphasia

Some people may also develop problems with movement:

- Motor neurone disease or MND
- Parkinsonism, of which two main types are seen called corticobasal syndrome or CBS, and progressive supranuclear palsy or PSP.

Diagnosis is usually made by a specialist and involves a clinical assessment and a series of tests.

Clinical assessment

The initial part of the diagnostic process is an assessment by a specialist. They will go through the symptoms that have been happening and ask about problems that run in the family. The specialist will also perform some tests of thinking, memory and language as well as a physical examination to look for signs of MND or parkinsonism.

It is important to have a partner, family member or close friend with you at this appointment.

Blood tests may be performed at this appointment to exclude some rare causes of dementia.

Genetic testing

In around a third of people FTD is due to a genetic problem with the main genes involved being called:

- Tau or MAPT
- Progranulin or GRN
- C9orf72

It is possible to test for problems in these genes at specialist centres by taking a blood test. This is done if there is a strong family history of dementia or if there are symptoms and signs suggestive of a genetic cause. It may take a number of months before a result is available.

Neuropsychology

Sometimes more detailed thinking, language and memory tests are performed by a neuropsychologist. This testing may take a couple of hours and provides the specialist with a more complete picture of the problems that are happening.

Brain imaging

Brain imaging should be performed to help make a diagnosis of FTD. This may be a CT scan but more commonly is a magnetic resonance imaging or MRI scan. This involves lying down in the scanner for around twenty minutes to half an hour. The images that are produced are then

looked at by a neuroradiologist, a doctor trained in looking at brain imaging, to see if there are signs of FTD.

Lumbar puncture

Also called a spinal tap, this is a medical procedure where cerebrospinal fluid or CSF is taken by inserting a needle into the lower part of the spine. The whole procedure takes about twenty minutes in total.

This is sometimes performed when trying to diagnose FTD and can be helpful to exclude other diagnoses.

EMG

If there is a suspicion that someone may have MND then a needle test of the electrical activity of the muscles is performed. This is called electromyography or EMG.

Apart from a genetic test there is no single test that diagnoses FTD. A specialist will gather all of the information from the different tests together to help them make a diagnosis. They will go through the results of the tests with you at a second appointment and tell you what they think the diagnosis is.

Sometimes the diagnosis is not completely clear at the first couple of appointments and it is only over time that the diagnosis becomes clear.

FACTSHEET 12

HOW IS FTD TREATED?

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

There is currently no cure for FTD but there are a number of important things that can help when caring for someone.

Drugs

There is some evidence that selective serotonin reuptake inhibitors or SSRIs may help improve behavioural symptoms. These are the drugs such as fluoxetine (Prozac).

The acetylcholinesterase inhibitors that are used in Alzheimer's disease have no evidence for any benefit in FTD and may make some behavioural symptoms worse. These are the drugs called donepezil (Aricept), rivastigmine (Exelon) and galantamine (Reminyl).

Neuroleptic drugs such as haloperidol, olanzapine (Zyprexa), risperidone (Risperdal) and quetiapine (Seroquel) have been used to treat behavioural symptoms but are associated with a significant risk of side-effects including the development of parkinsonism and deterioration in thinking.

In people with MND, the drug riluzole (Rilutek) is commonly used.

In parkinsonism levodopa (Sinemet, Madopar) or dopamine agonists such as ropinirole (Requip) or pramipexole (Mirapexin) may be used, although there tends to be limited benefit of these drugs on improving movement problems.

Environmental modifications

Sometimes modifications to the environment or other practical steps can be made to help deal with behavioural symptoms. Examples include:

Carrying a card to give to other people that briefly describes that the person's behaviour or judgment is affected by dementia – this can be useful when behaviour is disinhibited or inappropriate in public

Limiting access to credit cards if excessive spending is a problem

Limiting access to food if overeating becomes an issue

Speech and language therapists

In people with language problems, particularly those with progressive nonfluent aphasia, referral to a speech and language therapist may be helpful. In the early stages of the illness, the use of alternative communication methods such as electronic devices or picture books can be useful.

Genetic advice and counselling

Once someone has been diagnosed they may receive support from a variety of health and social care professionals.

This includes local health services such as GPs, community mental health teams and social workers, as well as the specialist hospital team.

Together, this team will help people and those who care for them to manage the challenges of FTD.

Support for patients

Once someone has been diagnosed they may receive support from a variety of health and social care professionals to help them and those who care for them manage the challenges of FTD. This includes local health services such as GPs, community mental health team and social workers.

Support for carers

It is important to that carers of people with FTD have enough support including respite when necessary.

Support groups can be helpful and include the following:

- Rare Dementia Support (UK)
  www.raredementiasupport.org
- Alzheimer's Society (UK)
  www.alzheimers.org.uk
- Association for Frontotemporal Degeneration (US)
  www.theaftd.org
- The Australian FTD Association (Australia)
  www.theaftd.org.au
- For links to other FTD support groups around the world
  www.worldftdunited.net
- PSP Association
  www.pspassociation.org.uk
- MND Association
  www.mndassociation.org

The future

Whilst there is currently no cure for FTD there are lots of research centres around the world working towards one.

Currently, people with FTD are often asked to take part in research studies looking at various aspects of the disorder including brain scanning and neuropsychology tests.

It is hoped that this research will lead to clinical drug trials over the next few years in FTD and its associated disorders.