What is a Rare Dementia?

Between 5% and 15% of people living with a dementia receive a diagnosis of a rare or young-onset dementia. The diagnosis of a rare dementia brings with it a set of unique and complex challenges which can include difficulties with vision, language, movement and behavioural changes.

30% of people living with a rare dementia first receive an incorrect diagnosis and there is a widespread lack of understanding about, and resources for people living with, rare dementias. By increasing awareness about these dementias we hope to empower the healthcare sector and communities to better support those affected by a rare dementia diagnosis.

This document will provide information about 6 of the rare dementias.

About Primary Progressive Aphasia (PPA)

The term Primary Progressive Aphasia (PPA) refers to a group of dementias, predominantly causing a loss of speech and language abilities. ‘Aphasia’ refers to a neurological language problem, ‘progressive’ being worse over time, and ‘primary’ as this condition is caused by brain tissue changes rather than an external cause.

Within the PPA group, three main patterns of language loss are recognised: Semantic Dementia (SD), Progressive Nonfluent Aphasia (PNFA) and Logopenic Aphasia (LPA). PNFA and SD fall within a larger group of brain disorders collectively called Frontotemporal Dementia (FTD – see below). LPA, by contrast, is an unusual form of Alzheimer’s Disease and is usually caused by the same proteins that cause Alzheimer’s disease.

Semantic Dementia (SD)

The first symptoms of Semantic Dementia (SD) are usually problems with language.

This can be:

- Difficulty finding the right word - often substituting another word or a vague term such as ‘thing’ instead of the specific word
- Loss of knowledge of what words mean or what objects are for
- Talking about things in a vague or ‘roundabout’ manner – referred to as ‘circumlocutory’ speech
- Difficulty understanding what other people are saying
- Problems with reading and writing

Progressive Non-Fluent Aphasia (PNFA)

The first symptoms of PNFA are usually difficulties producing speech.

This can be:

- Difficulty producing words - speech may be effortful and words may come out distorted. This is due to difficulties in the co-ordination of the movements of speech and is sometimes called ‘apraxia of speech’
- Difficulty organising words - the structure of the sentences may be affected with words being missed out and errors in grammar. Together these factors make the speech sound distorted, slow, hesitant and difficult to understand.

**Logopenic aphasia (LPA)**

The main symptoms of LPA are usually:

- Difficulties with finding the right word. Speech contains pauses, where the person stops what they are saying as they try to find the right word
- Slow and hesitant speech and the pronunciation of words may be affected
- Loss of other cognitive functions such as calculation and memory as the condition progresses

**About Posterior Cortical Atrophy (PCA)**

Posterior Cortical Atrophy (PCA) is a form of dementia predominantly affecting the processing of visual and spatial information. Common first signs and symptoms include difficulties with seeing what and where things are (e.g. when driving or reading).

PCA means ‘back of the brain shrinkage’ and it refers to the progressive loss of neural cells, starting in the occipital and parietal lobes. These changes can be caused by a number of different underlying disease processes. The majority of cases are caused by Alzheimer’s Disease and PCA is sometimes called the ‘visual variant of Alzheimer’s Disease’. PCA can also be caused by Dementia with Lewy bodies, corticobasal degeneration and other causes.

Common Symptoms include:

- Problems with reading
- Issues with recognition
- Managing co-ordination
- Visual problems
- Issues with judging distances
- Light sensitivity

**About Frontotemporal Dementia (FTD)**

Frontotemporal Dementia (FTD) is a form of dementia predominantly affecting behaviour and personality, with relatively few memory problems at the outset. It is caused mainly by a loss of cells in the frontal and temporal lobes of the brain. The main symptoms are a progressive change in personality and behaviour or progressive deterioration in language abilities.

FTD is subdivided into two types: Behavioural variant FTD (bvFTD) and Primary Progressive Aphasia (PPA - see above).
Behavioural variant FTD (bvFTD):

The first symptom of bvFTD is usually a change in personality or behaviour – the symptoms can come on very slowly and may not be noticed as abnormal at first. Quite often the person affected by bvFTD lacks any insight into these changes.

The symptoms can include:

- Loss of inhibitions or increased extroversion, for example making inappropriate remarks in public or being rude or impatient. The person may also become aggressive
- Excessive money spending
- Apathy or withdrawal from social activities
- Loss of empathy
- Changes in sexual behaviour: either more/less or inappropriate interest
- Being easily distracted
- Developing fixed routines or become obsessive about things, particularly time (‘clock watching’). Some people begin to hoard things
- Possible development of a sweet tooth or a preference for unusual foods. The person may also overeat leading to weight gain or drink excessive amounts of alcohol
- A compulsion to put objects in their mouth in the later stages

About Dementia with Lewy Bodies (DBL)

Dementia with Lewy Bodies (DBL) is characterised by the presence of proteins called ‘Lewy bodies’ in the brain. These proteins can also be found in people with Parkinson’s disease. Scientists still don’t know exactly how Lewy bodies cause dementia but they may lead to loss of connections between brain cells.

The symptoms that people with DBL will experience depend on where the Lewy bodies are found. When they are mostly found in the deeper parts of the brain, they lead to problems with movements and Parkinson’s disease. When they are found in the outer parts of the brain they tend to lead to Dementia with Lewy Bodies.

Common symptoms include:

- Problems with concentrating and staying alert
- Fluctuations in thinking and memory problems vary from day to day or even hour to hour
- Visual hallucinations – often of animals or people
- Slower of stiff movements. Some people have problems with falling
- Problems with bladder or bowel function
- Sleep disturbances

DBL is a progressive condition and symptoms generally become worse over time. The rate at which symptoms become worse varies from person to person.
About Familial Alzheimer’s Disease (FAD)

Familial Alzheimer’s disease (FAD) is an inherited form of Alzheimer’s disease, caused by a faulty gene - presenilin 1 (PSEN1), presenilin 2 (PSEN2) or amyloid precursor protein (APP) genes or APP duplications.

FAD is a rare form of Alzheimer’s Disease caused by specific genetic mutations that run within families. Familial Alzheimer’s Disease probably accounts for less than 1% of cases of Alzheimer’s Disease overall and generally affects people at a younger age than the more common, non-inherited form of Alzheimer’s Disease.

The age at which people develop symptoms varies considerably between different families but it is typically before the age of 65. In some families, individuals may be as young as their early 30’s when they become affected. Within a single family, people tend to develop the disease at broadly similar ages.

Individuals with familial Alzheimer’s Disease usually have a strong family history of the illness, which means that they know of cousins, aunts/uncles, parents and grandparents who were affected at a similar age. In some cases an individual may not know whether they have a family history of Alzheimer’s Disease, for example if they do not know their biological parents or if their parents died young.

About Familial Frontotemporal Demential (fFTD)

Familial Frontotemporal Dementia (fFTD) is an inherited form of Frontotemporal Dementia (FTD - see above), caused by a faulty gene which runs in families - by mutations in the tau, progranulin or C9ORF72 genes. Of the people diagnosed with FTD, a third is known to be caused by a genetic problem.

The largest group of people living with fFTD (20-40%) will have family members who have also had a diagnosis of either FTD or a related neurodegenerative disorder such as Alzheimer’s Disease or Motor Neuron Disease. However, the disease is not passed down from parent to child in these families. This type of dementia is termed familial as those affected may carry genetic risk variants that increase their risk of developing this dementia. However, this alone is not sufficient enough to cause FTD.

The inheritance pattern in familial dementias is complex and likely due to a combination of genes, lifestyle and environment. Some family members may carry the risk gene and remain completely unaffected. The genes that increase the risk of developing a dementia like fFTD are only just starting to be discovered and, because the link between these genes and dementia is not clear, there is currently no genetic testing available for risk genes.

In a smaller number of families living with fFTD (~10%), dementia is caused by a genetic fault. This type of dementia is called inherited dementia, and there is a clear family history of the disease being passed from parent to child. Specifically, every person affected in this group will have an affected parent, and each child of an affected person will have a 50% chance of developing fFTD. Several genes have been identified with faults that cause Frontotemporal Dementia.
What resources are available?

Rare Dementia Support [www.raredementiasupport.org](http://www.raredementiasupport.org)

Alzheimer’s Research UK: [https://www.alzheimersresearchuk.org/](https://www.alzheimersresearchuk.org/)

Motor Neuron Disease Association: [https://www.mndassociation.org/](https://www.mndassociation.org/)

Progressive Supranuclear Palsy Association: [https://pspassociation.org.uk/](https://pspassociation.org.uk/)

FTD Talk: [http://www.ftdtalk.org/](http://www.ftdtalk.org/)

Young Dementia UK: [https://www.youngdementiauk.org/](https://www.youngdementiauk.org/)

Parkinson’s UK [https://www.parkinsons.org.uk/](https://www.parkinsons.org.uk/)

Lewy Bodies UK [https://www.lewybody.org/](https://www.lewybody.org/)

About Rare Dementia Support (RDS)

*Rare Dementia Support* (RDS) is led by the UCL Dementia Research Centre. It holds regular support group meetings in London and regionally for each of the 6 rare dementias mentioned above, providing information, resources and peer-to-peer support for all those affected by these conditions (people living with a rare dementia, carers and loved ones). There are also separate meetings for carers and bereaved carers. Recordings of all meetings can be found on our website.

Newsletters, including the latest updates, events, personal stories, educational resources and research developments are available regularly throughout the year.

To receive newsletters and information about the support groups please follow the link: [https://tinyurl.com/raredementiasupport](https://tinyurl.com/raredementiasupport).

More information on rare dementias and Rare Dementia Support can be found at [raredementiasupport.org](http://raredementiasupport.org).

About The National Brain Appeal

*The National Brain Appeal* (Charity number 290173) raises money to fund Rare Dementia Support, holding and managing the *Rare Dementia Support Fund*.

Over the past three years The National Brain Appeal has committed to raise £150,000 per annum to provide support for the six rare dementias. Over 3,000 people are currently on support group databases - receiving emails and newsletters - and around 1,000 attend support meetings. Travel and accommodation bursaries are also available to help patients and carers attend the meetings.

In 2019 the charity is increasing its annual fundraising target to £250,000 and this target will continue to increase to £350,000 by 2022 to develop and extend the service, with the ultimate aim of providing everyone affected by a rare dementia with access to specialist information, support, education and contact with other people with a similar condition.