DementiaUK
Helping families face dementia

# Understanding genetic forms of dementia (familial dementia)

Genes carry hereditary information from one generation to the next, including characteristics like eye colour and height. It is estimated that parents pass around 20,000 different genes to their children.

In some families, a gene may be faulty (a 'genetic mutation'), and this can cause various health problems that are passed on from parents to children.

Research has identified some rare genetic mutations that can cause dementia. These are found on different genes depending on the form of dementia. If a person has one of these genetic mutations it means that they will eventually develop dementia. Every child they have will have a 50% chance of also inheriting the faulty gene and developing dementia in the future.

Genetic forms of dementia are more common in people under the age of 65, and it is thought that genetic mutations account for seven to 12% of all cases of young onset dementia.

Many people fear that dementia runs in the family and that if a parent or grandparent has it, they will develop it too, but in the vast majority of cases, it is not inherited. However, it is important to know your family history of dementia as you will be asked about this if you develop symptoms and seek an assessment and diagnosis.

## Which dementias can have a genetic link?

**Familial Alzheimer's disease (FAD)**
This is a rare form of Alzheimer's disease that is most common in people with young onset dementia (where symptoms develop before the age of 65). The genetic mutations tend to be on one of three genes: presenilin 1, presenilin 2 or amyloid precursor protein. Presenilin 1 (PSEN-1) abnormalities are the most common form, accounting for about 80% of all cases of FAD.

All three mutations cause a build-up of proteins in the brain called plaques. These disrupt the brain's normal processes and the ability of the brain cells to function effectively, leading to symptoms like memory problems; difficulties with cognition (thinking);

problems with speech and language; and changes in mood, personality and behaviour.

Please see Sources of support on p10 for more information on Alzheimer's disease.

**Familial frontotemporal dementia (fFTD)**

According to current research, 30-40% cases of frontotemporal dementia (FTD) are genetic. This is known as familial frontotemporal dementia (fFTD). It is most common in people aged 40 to 60, but even in this age group, the majority of cases of FTD are not inherited.

The most common form of fFTD is behavioural variant frontotemporal dementia (also known as bvFTD or Pick's disease). Symptoms of bvFTD include reduced motivation; inappropriate behaviour, eg making insensitive or suggestive comments or being overly familiar with strangers; reduced empathy; obsessive or repetitive behaviour; and changes in eating and drinking habits, eg binge eating or craving sweet foods. Memory is usually less affected in the early stages.

To date, there has been limited research into genetic mutations that can cause fFTD but they are thought to lead to a build-up

of a protein called tau in the brain, which can damage the brain cells and interfere with the communication between cells. Research is ongoing to understand more about fFTD.

Please see Sources of support on p10 for more information on frontotemporal dementia.

**Vascular dementia**
There is a rare form of hereditary vascular dementia called cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL). It affects about 1,000 people in the UK.

The genetic mutation causes a problem with a protein that is thought to be essential for the maintenance of blood vessels, including those that supply blood to the brain.

CADASIL causes strokes which can lead to limb weakness, slurred speech, confusion, changes in cognitive function, visual disturbances and migraine.

Please see Sources of support on p10 for more information on vascular dementia and CADASIL.

## How are genetic forms of dementia diagnosed?

People who have, or are suspected to have, a genetic form of dementia may want to know whether they are carrying a genetic mutation so they can plan for the future or be involved in research. In these cases, the GP should consider referring the person for genetic counselling and testing.

There are two types of genetic testing:

1. **Diagnostic genetic testing** – for people with a diagnosis of dementia who have a strong pattern of family inheritance. This confirms whether the person's form of dementia is genetic.
2. **Predictive genetic testing** – to detect genetic patterns and mutations in people who have a high risk of familial dementia, and assess the chances of them or their children developing dementia.

In both cases, a detailed background medical history needs to be established before genetic counselling and testing takes place. This should ideally span three

generations and look at the family history of:

- dementia
- serious mental illness (eg psychosis)
- neurological conditions (eg Parkinson's disease, motor neurone disease)

If any of these are present in any of the three generations, the person's age when they developed symptoms and, if applicable, their age at the time of death should be established.

Before having genetic testing, the person whose family history suggests there may be a familial type of dementia should receive counselling, along with their next of kin. This should include an explanation of the testing process and the potential implications of finding a genetic mutation.

Genetic counselling takes place over several months, both before and after the genetic testing. Both the person who is at risk of a genetic form of dementia and their next of kin – for example, their spouse/partner, parent, adult child or sibling – should consent to proceed with testing.

The person and their next of kin will be asked whether they want to know the results or would prefer them to be kept on file for access at a later date.

Finding a genetic mutation predicts that the person will develop dementia at some point in their life (unless they die of something else before the genetic mutation is activated) and can pass this to the next generation. Some people like to know this information as it can remove the uncertainty of not knowing whether they are carrying the genetic mutation.

If a person finds they do have a genetic mutation for dementia, they can take this into consideration when planning for the future or deciding whether to have children. Predictive testing can also enable the person and their family to be involved in research studies into the causes, potential cures, treatment and care of genetic forms of dementia.

However, there may be disadvantages in finding out that

there is a genetic mutation for dementia in the family, including the stress of knowing that it is a possibility and watching for signs that the process has started.

In addition, the knowledge that there is currently no known prevention or cure for familial dementia can lead to depression, grief and anger, and put added stress on a family's relationships – for example, if one family member discovers they have a genetic mutation for dementia and others do not.

It may also affect the person's decisions about whether to have children; or if they already have children, the possibility of them having inherited a genetic mutation may cause worry and distress about how they may be affected in the future.

After genetic counselling, many families decide not to proceed with predictive genetic testing; however, this is a very personal decision and everyone will have their own reasons for deciding for or against genetic testing.

## Can I prevent genetic forms of dementia?

Whilst genetic forms of dementia cannot be prevented, some research suggests the onset can be delayed by:

- having regular health checks with a GP (usually annually), eg blood pressure and cholesterol monitoring and blood tests to identify any deficiencies or a raised blood sugar level. Any abnormalities can then be treated and monitored
- having regular sight and hearing checks (usually annually) to identify and treat any problems
- increasing physical activity – guidelines recommend exercising moderately for 20 to 30 minutes per day, at least five days per week
- increasing the amount of vegetables, fruit, grains, beans, legumes, nuts, poultry and fish in the diet and cutting down on saturated fats, sugar and salt
- maintaining a healthy weight – a healthy midlife body mass index (BMI) is between 18.5 and 25.

You can calculate BMI at **nhs.uk/live-well/healthy-weight/bmi-calculator**

- stimulating the brain, eg by learning a new skill, reading, playing chess
- stopping smoking – please see **nhs.uk/live-well/quit-smoking** for advice
- keeping alcohol consumption within recommended limits – please see **nhs.uk/live-well/alcohol-advice/calculating-alcohol-units**
- ensuring you have social stimulation and opportunities to connect with other people
- avoiding prolonged daily stress
- aiming to have good, restorative sleep

## Managing genetic forms of dementia

Currently, there are no specific treatments for genetic forms of dementia but medication may be given for underlying conditions that could be

contributing to or increasing the risk of dementia. These are usually medications to treat high blood pressure, high cholesterol, heart problems or diabetes.

In cases of genetic forms of Alzheimer's disease, medications like donepezil, rivastigmine and galantamine may be effective. However, these medications are not effective for genetic forms of vascular and frontotemporal dementia, and may cause side effects such as nausea, diarrhoea, stomach pain, cramps and confusion. Please see Sources of support on p10 for information on medications and dementia.

People with a genetic form of dementia and their families often find that joining a specialist support group provides opportunities to connect with people who understand what they are going through and helps them manage their feelings and fears. Rare Dementia Support provides information on groups for people with genetic forms of dementia: please see Sources of support on p11.

If the person feels depressed or anxious about their diagnosis of dementia or the possibility of developing a genetic form of dementia in the future, the doctor may refer them for counselling. In some cases, the GP may prescribe medication to treat the symptoms of depression or anxiety. Please see Sources of support on p10 for information on dementia, anxiety and depression.

## Practical tips for managing genetic forms of dementia

These tips may help to manage the effects of the changes in the brain caused by a genetic form of dementia.

- People with dementia are often easily distracted, find it hard to concentrate and have short-term memory problems. To help them manage complex tasks, break them down into smaller steps
- Use reminders, pill boxes or automatic medication dispensers to ensure that any prescribed medication is taken regularly

- It is a good idea for the person with dementia to carry a card with details of their diagnosis and what sort of help they may need. This can be useful in situations in public where there may be misunderstandings or a need for extra support
- The Hidden Disabilities Sunflower scheme provides a range of cards, lanyards and other information that indicate to members of the public, shop staff and services such as public transport that the person has a hidden disability – please see Sources of support on p11 for information
- Peer and social support groups can provide an opportunity for people with genetic forms of dementia to share experiences and tips for preventing or managing potential difficulties. These may take place locally or nationally, and may be held face-to-face or online
- Noisy or crowded places could trigger changes in behaviour, so it may help to avoid these situations or provide support if they cannot be avoided
- Having a routine and regular activities can help the person to feel more relaxed and secure
- Look out for triggers for distressed behaviour such as being too hot or cold, noise, pain, misunderstanding, emotionally stressful situations, changes in routine, lack of activity or too much or too little stimulation
- Focus on what the person can still do rather than on areas they find difficult. Encourage them to keep up with activities they enjoy, eg photography, art, exercise, swimming, walking, volunteering, taking care of a pet
- If the person works, encourage them to tell their employer and colleagues about their diagnosis so support can be put in place to enable them to continue in their job if they want to. Please see Sources of support on p10 for information on employment and dementia

## Sources of support

To speak to a specialist dementia nurse about genetic forms of dementia or any other aspect of dementia, please call our Helpline on **0800 888 6678** (Monday to Friday 9am-9pm, Saturday and Sunday 9am-5pm) or email **helpline@dementiauk.org**

To book a phone or video call appointment with an Admiral Nurse, please visit **dementiauk.org/book-an-appointment**

### Dementia UK resources

**Alzheimer's disease**
**dementiauk.org/alzheimers-disease**

**Anxiety and depression**
**dementiauk.org/managing-anxiety-and-depression**

**CADASIL**
**dementiauk.org/cadasil**

**Employment and dementia**
**dementiauk.org/employment**

**Frontotemporal dementia**
**dementiauk.org/frontotemporal-dementia**

**Getting a diagnosis of dementia**
**dementiauk.org/getting-a-diagnosis-of-dementia**

**Medication for dementia**
**dementiauk.org/medication-management**

**Vascular dementia**
**dementiauk.org/vascular-dementia**

**Young onset dementia section**
**dementiauk.org/young-onset-dementia**

### Other resources

**Dementia Carers Count – free dementia training courses for family and friends**
**dementiacarers.org.uk**

**Dementia Engagement and Empowerment Project (DEEP) – a network of groups of people living with dementia**
**dementiavoices.org.uk**

**FTDtalk**
**ftdtalk.org**

**Genetic Alliance UK**
**geneticalliance.org.uk**

**Hidden Disabilities Sunflower scheme**
hiddendisabilitiesstore.com

**The Brain Charity**
thebraincharity.org.uk

**Rare Dementia Support**
raredementiasupport.org

**Young Dementia Network**
dementiauk.org/the-young-dementia-network

**Cambridge Stroke CADASIL website**
cambridgestroke.com/cadasil.php

The information in this leaflet is written and reviewed by dementia specialist Admiral Nurses. We hope you find it useful. If you have feedback, please email **feedback@dementiauk.org**

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Helpline: **0800 888 6678** or **helpline@dementiauk.org**
Virtual clinics: **dementiauk.org/book-an-appointment**

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