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 30,000 different genes to their children.

In some families, a gene may be faulty (known as 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 are likely to eventually develop dementia, although sometimes it can skip a generation or not show up until old age.

For most dementia genes, if a parent has a faulty gene, every child has a 50% chance of inheriting that gene. Most (but not all) of those with the faulty gene will develop dementia at some point in their life.

Genetic causes are more common in people with young onset dementia (where symptoms develop before the age of 65). Genetic mutations account for about one in 10 of all cases of young onset dementia, but this figure varies widely according to the type of 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 helpful to know your family history of dementia as you will be asked about it if you develop symptoms and seek an assessment.

Which dementias can have a genetic link?

Familial Alzheimer's disease (FAD)

Alzheimer's disease is usually not inherited, even if a parent or grandparent develops the condition in later life. But if symptoms

develop before the age of 65 (and particularly before the age of 55), it is sometimes due to a faulty gene. This is called familial Alzheimer's disease (FAD).

The mutations that cause FAD tend to occur on one of three genes:

- presenilin 1
- presenilin 2
- amyloid precursor protein

These mutations cause a build-up of proteins in the brain (called amyloid proteins) that form plaques. They 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
- changes in vision
- changes in mood, personality and behaviour

Please see Sources of support on p16 for more information on Alzheimer's disease.

Familial frontotemporal dementia (fFTD)
Frontotemporal dementia (FTD) is a rarer form of dementia. In some cases, it is caused by a genetic mutation. This is known as familial frontotemporal dementia (fFTD).

FTD is the most common form of dementia in people aged 40 to 60, but even in this age group, most cases are not inherited.

The most common form of fFTD is behavioural variant frontotemporal dementia (also known as bvFTD or Pick's disease). About a third of cases are due to a genetic mutation, typically on genes GRN, MAPT and C9orf72. These lead to a build-up of proteins that damage brain cells. Several clinical trials are underway to see if these faulty genes can be 'neutralised', but no treatments are yet proven to work.

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
- changes in eating habits eg binge eating or craving sweet foods
- muscle weakness and wasting in some mutations (C9orf72)

Memory is usually less affected in the early stages of FTD than in some other forms of dementia.

Please see Sources of support on p17 for more information on FTD.

Vascular dementia

Vascular dementia is a very common form of dementia that is often linked to other health conditions such as type 2 diabetes and high blood pressure. It is rare for vascular dementia to be caused by a gene mutation.

There is, however, 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 maintains blood vessels, including those that supply blood to the brain.

CADASIL can cause dementia, migraine and strokes. Symptoms include limb weakness, slurred speech, confusion, changes in cognitive function and visual disturbances.

Please see Sources of support on p16-17 for more information on vascular dementia and CADASIL.

How are genetic forms of dementia diagnosed?

There are two types of testing for genetic forms of dementia:

1. Diagnostic genetic testing: for people with a diagnosis of dementia. This confirms whether the person's dementia is caused by a genetic mutation.
2. Predictive genetic testing: to detect genetic patterns and mutations in people who are currently healthy, but are closely related to someone with a known genetic mutation.

People who have dementia may want to know more about the cause, including whether there is a possible genetic mutation. This can be determined through diagnostic genetic testing. Knowing if there is a mutation and if so, on which gene, can:

- help the person understand the cause of their condition
- make them aware of complications related to the genetic mutation to look out for
- guide decisions about treatment
- inform other family members about the potential risk of them developing a genetic form of dementia

- enable the person and their family to be involved in research, including clinical trials

Diagnostic genetic tests can be done by a specialist in a discipline such as neurology, psychiatry or genetics who is experienced in the management of dementia. GPs do not usually undertake genetic testing.

Predictive genetic tests are different. Only a specialist geneticist can arrange these, and only for people with a high risk of familial dementia with a known gene in the family. Some people find it helpful to know whether or not they have a mutation to remove the uncertainty, but the person should always have counselling before deciding whether to have predictive genetic tests.

For both forms of genetic testing, the person will be asked about their family history of dementia and related conditions, and their own medical history. The family history may include relatives with:

- dementia (even if their diagnosis was a different type)
- serious mental illness (eg psychosis, bipolar disorder)
- neurological conditions (eg Parkinson's, motor neurone disease)

If any of these are present, the person may be asked how old their family member was when they developed symptoms and, if applicable, their age when they died.

Before having genetic testing, the person should receive counselling, which may be one-to-one or with their next of kin. Counselling includes an explanation of the testing process and the potential implications of finding a genetic mutation.

Genetic counselling for predictive tests usually takes place over several months, before and after the actual test. The results of the test might have implications for other closely related family members, particularly the person's children and siblings, so they might want to discuss the decision with them first. However, they have the right to confidentiality as they consider and undergo testing.

For diagnostic and genetic tests, the person being tested (often with their next of kin) will be asked whether they want to know the results or would prefer them to be kept on file and be told at a later date, if they choose.

Pros and cons of genetic testing for dementia

Having a genetic mutation increases the risk that the person will develop dementia at some point in their life (unless they die of something else first). For most mutations, it means that they can pass this gene on to the next generation. But different genes can behave differently, and they vary in the age of onset of symptoms; the types of dementia they cause (even in the same family); and whether they skip a generation.

If a person finds out that they do have a genetic mutation for familial dementia, they can take this into consideration when planning for the future or deciding whether and how to have children (for example, if they would rather adopt a child). 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 familial 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. The knowledge that there is currently no known prevention or cure for dementia may lead to psychological effects, for example depression, grief and anger.

Knowing they have a genetic mutation for dementia could affect the person's decisions about whether to have children biologically. 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.

It can also put added stress on family relationships – for example, if one sibling discovers they have a genetic mutation for dementia and the other siblings do not.

After genetic counseling, some people 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 testing.

Can I prevent genetic forms of dementia?

If someone is carrying a mutation for dementia, it is not yet possible to prevent them developing a genetic form of the condition. However, research suggests the onset could be delayed by following a healthy lifestyle, for example:

- having regular health checks with a GP (usually annually), eg blood pressure, blood sugar and cholesterol monitoring, to identify any conditions that could be treated and monitored
- having regular sight and hearing checks to identify and treat any problems
- taking physical activity – guidelines recommend exercising moderately for 20 to 30 minutes per day, at least five days per week
- eating a healthy diet that includes plenty of vegetables, fruit, grains, beans, legumes, nuts, poultry and fish; and minimal amounts of saturated fats, sugar and salt. The NHS Eat well model is a useful guide: please see Sources of support on p18
- maintaining a healthy weight: please see Sources of support on p18 for NHS information

- stimulating the brain, for example through learning new skills like a language or craft; keeping mentally active through work, hobbies and sport; and connecting with other people
- not smoking – please see Sources of support on p18 for NHS information on stopping smoking
- keeping alcohol consumption within recommended limits – please see Sources of support on p18
- avoiding risky activities that may lead to head injury (for example cycling without a helmet)
- avoiding prolonged daily stress
- aiming to have good, regular sleep

Treating and managing genetic forms of dementia

Medication

Currently, the treatments for familial forms of dementia are the same as for non-familial (non-genetic) dementias.

For people with genetic forms of Alzheimer's disease, medications like donepezil, rivastigmine and galantamine may be effective. They do not cure or prevent dementia, but may slow down the progress and make the symptoms more manageable. However, these medications are not effective for genetic forms of vascular and frontotemporal dementia, and may cause side effects such as agitation, nausea, gastrointestinal problems, cramps and confusion. Please see Sources of support on p17 for information on medication and dementia.

Medication may also be prescribed for underlying conditions that increase the risk of dementia (eg high blood pressure, high cholesterol, heart problems or diabetes).

In some cases, medication can be used to manage some of the symptoms of dementia, for example:

- agitation
- impulsive behaviour
- apathy
- sleep problems
- psychosis
- incontinence
- depression
- anxiety

- Parkinson's-type symptoms
- complications of motor neurone disease

These medications are prescribed based on the person's individual circumstances. They cannot prevent, cure or slow the progression of dementia, but may make the symptoms more manageable.

Support

People with a genetic form of dementia and their families may find that joining a specialist peer 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 p18.

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 and/or to a support group. In some cases, the GP

may prescribe medication to treat the symptoms of depression or anxiety. Please see Sources of support on p16 for information on anxiety and depression in dementia.

Practical tips for managing genetic forms of dementia

People with dementia often find it hard to concentrate and have short-term memory problems. To help them manage complex tasks, break them down into smaller steps. You could use reminders (for example on a calendar or smart phone), pill boxes or automatic medication dispensers to ensure that any prescribed medication is taken regularly.

If the person with dementia struggles with communication, think about strategies that could help, for example using simple language and short sentences; avoiding complex instructions or open-ended questions; and using non-verbal methods like pictures or gestures to illustrate what you are saying. Please see Sources of support on p17 for tips for communication.

It is a good idea for the person with dementia to carry details of their diagnosis. This can be useful in situations where there may be misunderstandings or a need for extra support. The Hidden Disabilities sunflower scheme provides cards, lanyards and other resources to alert members of the public, shop staff and services such as public transport that the person has a hidden disability. The Young Dementia Network also produces free ID cards for people with young onset dementia – please see Sources of support on p18 for information.

Look out for, and try to avoid, triggers for distressed behaviour such as:

- being too hot or cold
- pain
- hunger and thirst
- tiredness
- noisy or crowded places
- misunderstandings or frustration
- emotionally stressful situations
- changes in routine
- lack of activity
- too much or too little stimulation

Having a routine and regular activities can help the person to feel relaxed and secure. Focus on what they can still do rather than on areas they find difficult. Support 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 p15 for information on employment and dementia.

Sources of support

If you are living with dementia or caring for someone with the condition, register for our free online sessions, 'Dementia: what next?' at **dementiauk.org/dementia-what-next**

To speak to a dementia specialist Admiral Nurse, call our free Helpline on **0800 888 6678** (Monday-Friday 9am-9pm, Saturday and Sunday 9am-5pm, every day except 25th December) or email **helpline@dementiauk.org**

If you prefer, you can book a phone or video call with an Admiral Nurse at a time to suit you: please visit **dementiauk.org/book**

Dementia UK resources

Alzheimer's disease
dementiauk.org/alzheimers-disease

Anxiety and dementia
dementiauk.org/managing-anxiety

Causes and risk factors of dementia
dementiauk.org/causes-and-risk-factors

Depression and dementia
dementiauk.org/managing-depression

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

Supporting children and adolescents when a parent has young onset dementia
dementiauk.org/supporting-children

Tips for communication
dementiauk.org/tips-for-communication

Vascular dementia
dementiauk.org/vascular-dementia

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

Other resources

The Brain Charity
thebraincharity.org.uk

Cambridge Stroke CADASIL website
cambridgestroke.com/cadasil.php

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

NHS alcohol advice
- nhs.uk/live-well/alcohol-advice

NHS Eat well guide
- nhs.uk/live-well/eat-well

NHS information on healthy weight
- nhs.uk/live-well/healthy-weight

NHS quit smoking advice
- nhs.uk/live-well/quit-smoking

Rare Dementia Support
- raredementiasupport.org

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

Young Dementia Network ID cards
- youngdementianetwork.org/young-onset-id

To speak to a dementia specialist Admiral Nurse about any aspect of dementia:

Contact our Helpline:
0800 888 6678 or **helpline@dementiauk.org**

Book a virtual appointment:
dementiauk.org/book

Our charity relies entirely on donations to fund our life-changing work. If you would like to donate to help us support more families:

- Call **0300 365 5500**
- Visit **dementiauk.org/donate**
- Scan **the QR code**

Thank you.

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