

Dementia in people with Down syndrome: What does the research tell us?

By Dr Liz Evans and Tanya Duckworth

Dementia is more common in people with Down syndrome than the general population. Here, we review research on why this is the case, along with recommendations for how families can support a loved one with dementia.

What is dementia?

The term *dementia* doesn't refer to one specific disease but a set of symptoms caused by a number of different brain disorders. Dementia results in a decline in a person's mental abilities—their capacity to think, reason, and remember.

Most people with dementia will experience changes like:

- declines in memory, with more recent information or events being harder to remember
- difficulty concentrating
- difficulty finding the correct words to say
- reduced capacity to plan, to pay attention, and poorer judgement
- feeling less motivated
- personality and behaviour changes.

When dementia occurs in people under the age of 65, it is called 'younger onset dementia'.

There are many different types of dementia with different patterns of symptoms. The most common form is Alzheimer's disease. People with Alzheimer's disease show progressive memory loss and a gradual decline in other skills. Their brains show changes in the form of a build-up of sticky plaques between the brain cells and tangles within the cells¹.

Dementia is not a normal part of ageing. Normal ageing does mean that the brain slows down, and it is common to find it harder to remember things as we age.

However, forgetting recent events and conversations, forgetting the names of family members, and losing skills we once had are not normal at any age.

But changes in a person's memory and thinking skills can also be caused by other medical conditions, many of which can be successfully treated. Examples include:

- a vitamin or mineral deficiency
- a mental health problem such as depression
- problems with sight or hearing
- a side effect of new medication, or even a change in how their body deals with existing medications
- an underactive thyroid (hypothyroidism)

Some of these conditions are more common in people with Down syndrome than the general population. So, any time a person is showing a decline in their thinking or memory, or changes to personality and behaviour, it is important to talk to their doctor about it straight away.

How common is dementia in people with Down syndrome?

Dementia, particularly Alzheimer's disease, is much more common in people with Down syndrome than the general population and it tends to occur at a younger age. Scientists believe this is because a gene on chromosome 21 called the amyloid precursor protein (APP) gene plays a major role in the brain changes associated with Alzheimer's disease. Genes are a code for proteins, and because most people with Down syndrome have three copies of this gene, they get more of its protein¹.

Different studies have found very different rates of dementia in people with Down syndrome, ranging from under 10% up to 49 years of age, to around 30% for those in their 50s, and over 50% for those aged over 60². One recent study found a rate of just over 50% in those over 60 years³, but another recent study found a rate above 80% in those over 65⁴. The average age for diagnosis is in the mid 50s⁴.

Research suggests that virtually all people with Down syndrome have a build-up of amyloid plaques in their brain by about the age of 40¹. Yet for most, the outward



symptoms of dementia do not start for some years after this and some studies report people who live into old age without developing symptoms.³ So, it is not inevitable that a person with Down syndrome will get dementia but, due to the increased risk, it is still likely.

What are the signs of dementia in people with Down syndrome?

In people without Down syndrome, the earliest signs of Alzheimer's disease are usually memory problems. **But in people with Down syndrome, the first signs noticed by carers are more likely to be changes in behaviour and personality**, such as increased stubbornness and behaving inappropriately. Other early signs include difficulty paying attention and lower ability to plan, solve problems, and make judgements^{5,6}.

Other changes may include^{5,6}:

- apathy
- social withdrawal
- increased dependency
- confusion
- prolonged sadness
- fearfulness
- repetitive speech
- getting lost or disoriented in familiar places
- irritability or aggression
- seizures for the first time in adulthood.

What is the latest research on dementia in Down syndrome focused on?

Scientists are working in a number of areas to further knowledge about the link between Down syndrome and dementia. Two important areas of current research are regarding diagnosis, and possible future treatments.

Our own research team is conducting the *Successful Ageing in Intellectual Disability (SAGE-ID) Study*. One of the aims of that study is to compare different screening tools and assessments that may be suitable for people with intellectual disability, including those with Down syndrome. A further aim is to look at the factors associated with a higher risk of dementia in this group. People with intellectual disability aged over 40 can participate in the study, including those with or without dementia. This is to ensure a good mix of those who are healthy and those experiencing declines.

Other researchers are looking at different biological markers that might be able to identify the brain changes associated with Alzheimer's disease much earlier—even before cognitive symptoms begin. Examples include protein changes in a person's blood, new types of brain

scans, and measuring brain waves through EEG. If such measures could reliably detect brain changes earlier, then this could one day aid in directing specific therapies during the window before symptoms start^{1,7}. It could also help researchers as they try to develop and test future therapies focused on preventing dementia.

Certain medications can help to slow the rate of cognitive decline in some people who have dementia. However, studies with people with Down syndrome have found inconsistent results about whether these medications are effective, though some case studies suggest they may be for some people^{8,9}. However, people with Down syndrome may have an increased risk of side effects from these medications⁹.

Newer research is trying to develop future treatments that could prevent or alter the course of Alzheimer's disease⁷, not just address the symptoms. Much of the research on drugs and neurotransmitters (brain chemicals) is done initially using mice. The safety and usefulness for humans then needs to be established.

A handful of studies have also looked at whether antioxidants could prevent or slow Alzheimer's disease in people with Down syndrome¹⁰. So far, the results have not found that antioxidants worked to prevent decline⁸. Instead, results have pointed to the highly complex nature of the brain changes that lead to cognitive decline in people with Down syndrome. Much more research will be needed before scientists can identify specific supplements that may reduce dementia risk in people with Down syndrome.

Are there factors that increase—or decrease—the chances that a person with Down syndrome will develop dementia?

There is only a small amount of research about risk factors specific to people with Down syndrome. A handful of case studies suggest that people with atypical forms of Down syndrome may have a lower risk of developing Alzheimer's disease¹¹. Other studies have looked at the role of particular genes known to influence risk in the general population, but results are not always consistent across studies. Results regarding the potential influence of gender, hormones, and level of intellectual disability have also varied between studies.

However, much of what is known about dementia in the general population could also apply to those with Down syndrome. There is a considerable amount of evidence from the general population to support the protective effects of a healthy lifestyle. Research regarding people with Down syndrome is lacking but the World Health Organisation¹² recommends that people with intellectual disabilities should focus on similar targets.



A healthy lifestyle aiming to reduce dementia risk would include good nutrition, regular exercise, and not smoking¹²⁻¹⁴. People (in the general population) who regularly do moderate-intensity exercise have a lower risk of dementia. They also have a higher brain volume in areas related to memory, planning, and learning. The Mediterranean diet has also been found to reduce dementia risk in the general population as has staying socially active and engaging in stimulating activities for leisure, work, or education¹³. One of the most important elements of a healthy lifestyle is preventative health care including regular medical check-ups. Good physical and mental health throughout life is associated with a lower dementia risk in the general population¹³. People with intellectual disabilities often have undiagnosed or untreated health conditions which could be treated. Sensory problems and physical disabilities can also compound their health and quality of life.

In the general population, cardiovascular disease is a particularly important risk factor for dementia¹⁵. In general, people with Down syndrome have an overall lower risk of cardiovascular disease than the general population. However, it is reasonable to assume that for those people with Down syndrome who do have risk factors for cardiovascular disease, these factors would increase the risk of dementia. Such risk factors include a family history of heart disease and stroke, having diabetes, low levels of physical activity, a diet high in saturated fats, and smoking¹⁶.

Obstructive sleep apnoea is also known to increase the risk of dementia in the general population and it is very common in people with Down syndrome. It is possible that this could be an important additional risk factor for people with Down syndrome¹⁵. Medical management of obstructive sleep apnoea is based on an individual sleep study.

How can families identify the early stages of dementia and differentiate those from mental illness or other problems?

Diagnosing dementia in people with Down syndrome can be difficult. The standard tools for assessing cognitive function in the general population are not suitable when someone has an intellectual disability. What is needed is to compare the person's functioning to what it was before symptoms began⁹—but the person's typical level of function may not be well documented. As a result, health professionals rely on information provided by family, carers and other people who know the person well, to help come to an accurate diagnosis⁶. So it is important for the people close to the person with Down syndrome to



● Flying Kites at Challenge weekend

know the early signs of dementia and to consult a doctor about any changes observed or any other concerns. The earliest noticeable signs in people with Down syndrome may be behavioural or personality changes. If a person with Down syndrome consults a doctor when these changes are observed, then memory and other cognitive testing can be carried out at regular intervals to help to determine if decline is also occurring⁵.

There are tools available such as the Early Detection Screen for Dementia recommended by the National Task Group in the US. This is a tool that can help you to track your loved one's skills and any changes in their functioning time. At present, the tool does not provide a cut-off score: rather, it is designed to facilitate talking about any observed changes with a health professional.

While families and carers are critical to recognising changes in their loved one, consulting a doctor is essential to determining whether those changes might be dementia or something else. There are other conditions that may look the same as dementia, many of which can be tested for and treated.



What can parents/carers do to prepare for the management of dementia in their loved ones?

If your loved one develops dementia, the keys to supporting them will be early planning and working well with their doctor and other professionals. So encourage your loved one to find an attentive doctor they feel comfortable with, and to continue to see that doctor for annual health checks.

Early planning for any transitions begins with getting a diagnosis as early as possible. A baseline assessment of their skills when healthy is helpful. Use the free screening tool and, if resources permit, arrange an assessment with a psychologist or psychiatrist. Current recommendations are that people with Down syndrome have a cognitive assessment around age 30 to establish their normal level of functioning before declines begin and again at age 40¹⁷. But if they start to show declines, the assessment could be repeated annually⁵.

Build as much of a support network as possible around the person with Down syndrome. If dementia is diagnosed, talk with the person with Down syndrome about who is in their life and who they would like to invite to be involved in their care. Wherever possible, include the person as early as possible in the planning process. This may include discussions of end-of-life care¹⁸. Support your loved one to understand their diagnosis so that they may participate in this planning. An easy-read fact sheet with pictures can help (see the resources links below). Find out what your loved one's preferences are regarding care options, end-of-life planning, and what is important to them for their care¹⁸.

Families can facilitate holding onto items, such as photos and holiday souvenirs, which may one day serve a purpose in a memory box or life story. These are tools which can assist someone with dementia who is beginning to lose their memory. They can also aid communication between a person with dementia and others, and may help paid workers to understand the person better¹⁸.

Look into available services. People with Down syndrome have the right to access mainstream health services and aged care services. Those with younger onset dementia (before the age of 65 years) can also access aged care services if they have a diagnosis or suspected dementia. The National Younger Onset Dementia Keyworker Program can be accessed before a formal diagnosis is made. Of course, people with Down syndrome and dementia also remain eligible for disability-related

supports. A range of allied health professionals may be involved in the care of someone with intellectual disability and dementia to promote their wellbeing.

As dementia progresses, the care goal needs to shift from supporting independence towards providing care and eventually palliative care^{19,20}. Many people with Down syndrome and dementia may want to remain where they are living and their families may want this too²¹. However, if and when their care requirements can no longer be met in their current place, options will include transfer to an aged care facility or another disability service. Long-term planning for such transitions is helpful.

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You can find the list of references for this article on page 34.

Resources

An easy read factsheet is available from the Alzheimer's Society (UK) here: www.alzheimers.org.uk/site/scripts/download_info.php?downloadID=1092

The screening tool recommended by the US National Task Group is available from this site: aadmd.org/ntg/screening

Dementia Australia has made a video about dementia in people with intellectual disability. www.dementia.org.au/videos/collections?playlist=IntellectualDisability

If you would like further information, or would like to talk to us about the SAge-ID study, please phone Tanya or Liz on (02) 9931 9160 or email us at sageid@unsw.edu.au.

