

Down syndrome and Alzheimer's disease

This help sheet explains what is currently known about the link between Down syndrome and Alzheimer's disease and provides information about where to find additional support.

What is Down syndrome?

Down syndrome is a condition in which a person is born with a third copy of chromosome 21, instead of the usual two copies. This genetic abnormality can cause developmental problems and health issues and can affect learning, language and memory.

What is Alzheimer's disease?

Alzheimer's disease is a brain disease that causes dementia, resulting in impaired memory, thinking and behaviour. It is the most common form of dementia. Alzheimer's disease is characterised by specific changes in the brain. There is an abnormal build-up of a protein called beta amyloid, which forms 'plaques' outside the brain cells. Inside the brain cells, another protein called tau builds up into 'tangles'. These abnormal protein accumulations disrupt messages within the brain because they damage connections between brain cells. The brain cells eventually die and brain volume shrinks.

The brain degeneration that occurs in Alzheimer's disease affects memory, thinking skills, emotions, behaviour and mood. As a result, a person's ability to carry out daily activities becomes impaired. As the disease progresses, symptoms worsen.

The link between Down syndrome and Alzheimer's disease

Studies show that by the age of 40, almost all people with Down syndrome will have the changes in the brain associated with Alzheimer's disease. Because these changes build up slowly, they may not have the symptoms of dementia at this age, but they may develop in the future. Not everyone with Down syndrome develops dementia however.

The gene responsible for the amyloid precursor protein (APP) is located on chromosome 21. When APP is broken down, beta amyloid forms. Beta amyloid clumps together into the brain plaques that characterise Alzheimer's disease. Because people with Down syndrome have an extra copy of chromosome 21, they make 1.5 times as much APP as other people, and make more beta amyloid. This appears to cause earlier appearance of the brain changes typical of Alzheimer's disease.

People with Down syndrome therefore have a much higher chance of developing Alzheimer's disease, and at a much earlier age, compared to the general population. Most studies report that around 50% of people with Down syndrome will develop Alzheimer's disease by the age of 60 years, and that the average onset age is in the early to mid 50s.

The early signs of dementia

Those with Down syndrome may show different symptoms in the early stages, or symptoms may be a further deterioration of pre-existing difficulties and thus it may be harder to recognise. Common early signs include:

- Decreased ability to accomplish tasks of daily living
- Deterioration in short term memory
- Increased apathy and inactivity
- Reduced interest in being sociable and a reduction in spontaneous communication and communication skills
- Increased difficulties understanding language
- Disorientation and confusion
- Changes in night time sleep patterns
- Sadness, fearfulness or increased anxiety
- Restlessness

If a person with Down syndrome exhibits some of these changes from their normal behaviour, they should have a thorough assessment to determine if the changes suggest dementia. The possibility that they are the result of other conditions also needs to be investigated. Treatment of these other conditions can lead to great improvements in the person's functioning and well being. It is very important to get an early diagnosis so that the person can be given appropriate support and access to services.

Diagnosing Alzheimer's disease in someone with Down syndrome

The process of making a diagnosis of Alzheimer's disease follows a similar process for everyone. It will include tests of thinking and memory, physical examinations and investigations (blood tests and brain scans) and the collection of a detailed history of progressive changes over time in cognitive and daily living skills from the person themselves and/or family members or friends. Tests will also be completed to rule out other conditions or possible causes of decline in functioning.

The difference for people with Down syndrome is that they have pre-existing difficulties with thinking, memory and daily living skills. Therefore it is important for the clinician to have a very clear picture of the person's abilities before any change was noticed and how the person's abilities have progressively changed. Reports from family and carers about the person's medical history can help to separate pre-existing disabilities from the symptoms of Alzheimer's disease. Documenting baseline functioning before age 35 can be helpful and on-going evaluation of intellectual, behavioural and social functioning may assist in picking up early changes.

If Alzheimer's disease is diagnosed

At present there is no cure for Alzheimer's disease and no treatment that can stop the disease progressing. However, there are medications available that may help stabilise or slow the decline in memory and thinking abilities for a time. People with Down syndrome may require smaller doses and may be more likely to develop side effects. Drugs may also be prescribed for secondary symptoms such as agitation or depression, or to help the person sleep better. It is recommended you speak to your specialist further about the best options for you.

Someone with Down syndrome that receives a diagnosis of Alzheimer's disease may not understand what that means for them, however information and specific supports are available to help with understanding changes and developing helpful strategies for the person with the diagnosis and their families and carers.

Activities and communication strategies may need to be modified to take into account the person's deteriorating memory and thinking skills. Some people with Alzheimer's disease may develop behaviours that cause concern to families and carers. Specialised support and advice is available to help manage these behaviours.

Support services are available for the person with Down syndrome and Alzheimer's disease, their family and carers. This support can make a difference to living well with these conditions. For people with Down syndrome who live in a residential setting, Dementia Australia can assist residential service providers to understand and manage the changes produced by Alzheimer's disease. Dementia Australia provides support, information, education and counselling for all people living with dementia, their families and carers.

Further information on Down syndrome

Down Syndrome Australia is a network of state-based organisations providing support, encouragement, information and resources to people with Down syndrome, their families and the broader community. Contact the Down Syndrome Association for further information at downsyndrome.org.au

Down syndrome and Alzheimer's disease is a booklet providing more comprehensive information. It was produced by Dementia Australia, Down Syndrome Australia and the Centre for Developmental Disability Health Victoria, and is available from cddh.monashhealth.org

Further Information

Dementia Australia offers support, information, education and counselling. Contact the National Dementia Helpline on **1800 100 500**, or visit our website at dementia.org.au



For language assistance phone the Translating and Interpreting Service on **131 450**