

Down syndrome and Alzheimer's disease

1.18

ABOUT DEMENTIA

Research has established a link between Down syndrome and Alzheimer's disease. This Help Sheet explains what is currently known about this link, and discusses the diagnosis of Alzheimer's disease in people with Down syndrome and some information about where to find additional support.

What is Down syndrome?

Down syndrome is a genetic disorder caused when, due to a biological error at the time of conception, a person has 3 copies of chromosome 21 in every cell body instead of the usual pair, so upsetting the gene balance. This extra gene material causes a collection of characteristics which can result in some degree of developmental delay and some common physical traits.

What is Alzheimer's disease?

Alzheimer's disease is a physical condition that affects the brain, resulting in impaired memory, thinking and behaviour. It is the most common form of dementia and accounts for 50% - 70% of all cases.

The link between Down syndrome and Alzheimer's disease

Studies show that by the age of 40, almost 100% of people with Down syndrome who die have the changes in the brain associated with Alzheimer's disease.

Amyloid precursor protein (APP), which is the abnormal breakdown that yields the toxic amyloid protein that forms plaques in the brain and probably damages brain cells and their connections, is coded for chromosome 21. Because people with Down syndrome have an extra copy of chromosome 21, they make 1.5 times as much APP as other people, and this seems to result in an excess tendency for the abnormal amyloid breakdown product to build up. This appears to cause earlier appearance of the brain changes typical of Alzheimer's disease.

However, a significant number of people with Down syndrome are older than 40 and show no signs of having Alzheimer's disease. It is not currently understood why changes to the brain that are typical of Alzheimer's disease do not necessarily produce the condition in people with Down syndrome.

Diagnosing Alzheimer's disease

Making a diagnosis of Alzheimer's disease can be difficult when the person has Down syndrome for a number of reasons:

- People with Down syndrome are susceptible to a number of reversible conditions that can be mistaken for Alzheimer's disease. These include hypothyroidism and depression. The side effects of some medications can also mimic Alzheimer's disease
- The usual skill tests used for diagnosis do not take into account the existing problems of a person with Down syndrome
- The limited communication skills of some people with Down syndrome may affect the assessment

Only a doctor can make a diagnosis of Alzheimer's disease. Making the diagnosis in a person with Down syndrome is usually based on excluding other possible causes of symptoms. 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.

If Alzheimer's disease is diagnosed

At present there is no cure for Alzheimer's disease. However some medications and alternative treatments have been found to reduce symptoms. Cholinesterase inhibitors such as Donepezil (Aricept), Rivastigmine (Exelon) and Galantamine (Reminyl) can be helpful treatments in some patients with Alzheimer's disease. These drugs though have not been systematically studied in individuals who have Down syndrome.

Drugs can be prescribed for secondary symptoms such as restlessness or to help the person sleep better.

Activities may need to be modified to take into account the person's deteriorating memory. Some people with Alzheimer's disease may develop behaviours that cause concern to families and carers. The principles of managing these behaviours are different from those which families and carers may be accustomed to, so specialised advice should always be sought about any changed 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 managing these conditions. Many people with Down syndrome live in a residential setting and Alzheimer's Australia can assist residential service providers to understand and manage the changes produced by Alzheimer's disease. Alzheimer's Australia provides support, information, education and counselling for people affected by dementia, including up to date information about drug treatments and research into dementia.

Remember

Families, friends and carers continue to play an important part in the lives of people with Down syndrome and Alzheimer's disease. They provide valuable links to past experiences, and enable the person to continue to be a loved and valued member of a family and circle of friends.

This Help Sheet was developed in collaboration with the Down Syndrome Association of Victoria.

Further Information

The Down Syndrome Association is a non-profit, self-help group established to bring people of like needs together, provide understanding support, information, resources and encouragement to people with Down syndrome and their families and to promote in every way possible improvements in the life of people with Down syndrome. It is a well used community resource and shares information and resources with other people and services. Contact the Down Syndrome Association in your State or Territory.

Alzheimer's Australia offers support, information, education and counselling. Contact the the National Dementia Helpline on **1800 100 500**.

For further information and to view other Help Sheets visit our web site at **www.alzheimers.org.au**

For up to date and detailed information about drug treatments on the web site go to the Update Sheets section.