Dementia in people with Down syndrome

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Dementia is a general term used to describe a group of symptoms caused by disorders affecting the brain. These disorders affect thinking, behaviour and the ability to perform activities of daily living. The most common form of dementia is Alzheimer's disease, a progressive disorder caused by a build-up of abnormal protein in the brain and a consequent loss of connections between nerve cells (neurons) in the brain.

Alzheimer's dementia is particularly common in people with Down syndrome. People with Down syndrome develop 'early onset' of this disease, which is defined as the beginning of symptoms before the age of 65. Holland, et al (2000) estimated that up to 75% of adults with Down syndrome develop Alzheimer's disease before the age of 65. This is therefore a problem associated with middle age for people with Down syndrome.

As family members and significant caregivers, it is important to be aware of the possibility of Alzheimer's disease for people with Down syndrome. It is also important to know (a) the early warning signs; (b) the ways of diagnosing Alzheimer's disease; and (c) what other conditions may share the same symptoms as Alzheimer's disease. Through this understanding we can facilitate early detection of the disease, and early access to the medical and social supports that may be needed. Concomitantly, we may facilitate the early detection of other treatable conditions which, while they may present like dementia, can be properly identified and treated correctly.

Early warning signs

Alzheimer's disease is characterized by a progressive, but not rapid, decline in skills. In people with Down syndrome the disease usually manifests through changes in personality and behaviour, and declining abilities. In the general population deterioration in episodic memory is usually the initial sign of dementia. (Beaumont, 2011; Ball, et al, 2008).

1. **Changes in personality and behaviour:**

These include extremes in behaviour, such as irritability, lashing out or self-abuse which were not previously evident, or deficits in behaviour, such as apathy, or withdrawal which, again, were not previously evident.

Example: Michael used to be compliant and helpful when it came to helping with household tasks, now he swears when asked to sweep the floor.

2. Decline in skills:

Executive skills (organising and completing complex tasks)

Example: Marion no longer boils water for her tea, she used to make a nice hot cuppa, but now she pours cold water from the tap directly onto her tea bag.

Language skills (using familiar words, complex sentences, comprehension, reading and writing)

Example: Reece used to sign his name, but now he pens random letters and is unsure of when to stop writing.

Recognition skills (knowing who (people), what (objects), or where (places) something is)

Example: Pete no longer uses his ipod even though it used to be his most prized possession and he wouldn't leave the house without it. He still loves music, but he can't quite figure out what his ipod is anymore. He fiddles with it sometimes, but leaves it in his room now.

Visual-spatial skills (sense of direction, artwork, negotiation of different floor types and stairs)

Example: Frank can no longer walk over the black and white tiles at the shopping centre – he is scared he is going to fall.

Learnt motor skills (folding, swallowing, walking)

Example: Don asks you to do up his shoes, even though he's been doing them independently for years. When you refuse he gets frustrated and knots his laces.

Memory loss which can be defined as progressive and frequent.

Example: Cate no longer recognises her baby niece, but is excited every time she hears about her birth as though she is learning about it for the first time.

Onset of **seizures**, or worsening of pre-existing seizures, may also be an early indication of developing Alzheimer's disease.

How Alzheimer's disease is diagnosed

When approaching a medical practitioner for a diagnostic evaluation, there are a number of steps that will be undertaken. The doctor will be looking for changes over time, and therefore a diagnosis is unlikely to be made on the first visit. The following are a list of the assessments routinely undertaken when diagnosing – or excluding – Alzheimer's disease:

1. Detailed medical history

The doctor will need to know about the person's past medical history and any current medical issues, as well as how the person has changed over time. Make sure that a carer who is familiar with the person with Down syndrome attends the medical appointment – ie, someone with a longstanding relationship with the person who has a good knowledge of their medical history. Take all previous health/clinical notes, list of current medications, and any notes about behaviours or other concerns, to the appointment with you.

2. Record of baseline skills

The medical practitioner will need to know what the person with Down syndrome was capable of doing before the onset of symptoms. S/he will be particularly interested in how the person/s behaviour, mood, personality, memory and skills have changed over time. The person's means of communication, interests and hobbies, expressions of happiness or sadness, interests and hobbies, eating habits, mobility, ability to learn new skills, self care and activities of daily living, memory, IQ, literacy and numeracy are all important in this assessment.

3. Physical examination

The medical practitioner will want to do a full physical examination, and so it is helpful if the person is prepared in advance for the need to get partially undressed for this part of the consultation.

4. Investigations

There are a range of blood, and possibly urine, tests that will be required, most to exclude other causes of changes in behaviour and skills. Preparing the person ahead of time in whatever way has worked best in the past for blood tests is likely to be beneficial.

The doctor may want to explore medical concerns, or causes of the changes, through others tests including XRays, EEG or CT scan. The process for these should be discussed in detail so parents/carers can prepare the person appropriately and support them through the procedure.

5. Mental status tests

These tests will include tests of memory, language, and attention. For these tests to be effective, a thorough understanding of baseline skills is important (see point 2 above).

6. Psychiatric assessment

A full psychiatric assessment may be required. This includes questions about the person's mood, thoughts and behaviours and seeks to identify any symptoms such as hearing voices. This assessment is important in identifying potentially treatable conditions, and to better understand the person's current experience.

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What else could it be?

There are a large number of health conditions which need to be considered before a diagnosis of Alzheimer's dementia can be made. All can cause a decline in cognitive function and skills similar to those seen in Alzheimer's; however many are treatable and so it is vital they are not overlooked. These include:

- Vascular dementia
- Head injury
- Delirium
- Depression
- Medication side effects
- Vitamin B12 deficiency
- Thyroid dysfunction
- Heart failure or abnormal heart rhythm
- Sleep apnoea
- Hearing/vision impairment
- Undiagnosed pain
- Environmental changes

Key message

If you notice any of the early warning signs listed here, it is important to consult a medical practitioner immediately. The changes may be explained through the diagnosis of a treatable condition which, once identified, can be effectively managed. If a diagnosis of dementia is made, this knowledge will enable you to access support and information to assist a person with Down syndrome to continue to achieve a high quality of life.

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References

Ball, S, Holland, A, Treppner, P, Watson, P, & Huppert, F (2008) **'Executive dysfunction and its association with personality and behaviour changes in the development of Alzheimer's disease in adults with Down syndrome and mild to moderate learning disabilities'** *British Journal of Clinical Psychology,* 47(1), 1 - 29

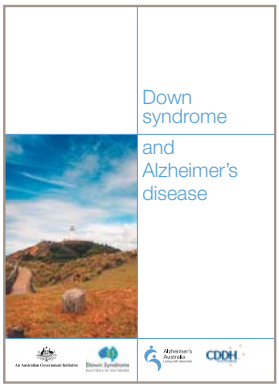
Beaumont, M (2011) **'Caring for people with Down syndrome and Alzheimer's disease in the early stages of assessment'** *Learning Disability Practice,* 14(4), 33 - 52

Holland, A, Hon, J, Huppert, F, & Stevens, F (2000) **'Incidence and course of dementia in people with Down syndrome: Findings from a population based study'** *Journal of Intellectual Disability Research,* 44(2), 138 - 146

For more information about Alzheimer's disease:

Down syndrome and Alzheimer's disease can be downloaded from the Down Syndrome Victoria website, or from the Centre for Developmental Disability Health Victoria (CDDHV) website:

www.cddh.monash.org/products-resources.html#downsynd



Down's syndrome and Alzheimer's disease. A guide for parents and carers from the Down's Syndrome Association (UK) can be downloaded at:

www.downs-syndrome.org.uk/images/documents/1090/DS_and_Alzheimers.pdf