Down's Syndrome and Alzheimer's Disease

A Guide for Parents and Carers

A DOWN'S SYNDROME ASSOCIATION PUBLICATION

INTRODUCTION

The life expectancy of people with Down's syndrome has, like that of the rest of the population, improved significantly and many people with Down's syndrome are able to live healthy adult lives, free from concerns about serious illness or additional disability.

Growing older is, however, associated for all us with an increased risk of developing certain illnesses, and it is therefore important to be aware of what can be done to prevent such problems arising, to recognise these problems when they do arise, and to then ensure that the right treatment and necessary support is available. Knowledge is important, as it is the key to proper treatment and support and can help you plan for the future, both as a person with Down's syndrome and as a family.

This booklet reviews three broad areas of potential concern. Where appropriate, reference is made to other organisations and sources of information. The three areas are as follows:-

1. life expectancy and ageing in people with Down's syndrome;
2. the specific association between Down's syndrome and the risk of Alzheimer's disease in later life; and
3. treatment and support strategies for later life.

Down's syndrome is associated with the inheritance of three, rather than the normal two, copies of chromosome 21. At first sight it might seem odd that there should be concern about later life in those that have a chromosome abnormality present from conception, and specifically, why there should be concerns about Alzheimer's disease, a cause of dementia predominantly affecting the elderly. It is now clear that genetic abnormalities present from conception can have an adverse influence that may only become apparent in later life. It is by no means inevitable, however, that all people with Down's syndrome will go on to develop Alzheimer's disease. The incidence of the disease is no higher than in the general population, although it is thought to occur 30-40 years earlier.

Life expectancy and ageing in people with Down's syndrome

The average life expectancy of people in the general population in Europe and North America has increased over the twentieth century, and is now in the mid-seventies. Women still tend to live longer than men, and there is some variation between affluent countries, possibly related to diet and lifestyle.

For people with Down's syndrome, whilst life expectancy has also increased dramatically over this period to an average of fifty years, it is still a third less than the average life expectancy for people without Down's syndrome. The possible reasons for the improvement in life expectancy are as follows: -

1. Better nutrition and living standards (as for the general population)

2. Effective prevention and/or treatment of infectious illness and other serious diseases, such as pneumonia, measles, etc.

3. Improved surgical and medical treatment of congenital heart disease and its consequences

The reasons why life expectancy for people with Down's syndrome is still lower than for those in the general population is uncertain. Here are two possible explanations: -

1. People with Down's syndrome "age prematurely" and therefore life expectancy is reduced

2. Later life for people with Down's syndrome is associated with an increased risk of illnesses that are associated with an increased mortality.

Each of these two possibilities will now be explored in more detail.

Premature ageing in people with Down's syndrome

For everyone a combination of genetic and lifestyle factors, together with chance occurrences, have an impact on how long we live. Some families will be known for their longevity. In these families it is likely that they have a particular genetic makeup and/or lifestyle that helps each of them to remain more resilient well into later life.
Since Down's syndrome was first described, it has been argued that people with the syndrome "appear to age faster or sooner". This has not systematically been shown to be the case. However, it is certainly feasible that having three copies of chromosome 21 in some way alters the way in which the body ages. Why might this be important? If it is true that the ageing process is speeded up or starts earlier then it is likely that some of the effects of ageing will be present earlier in a person's life and that the risks for illnesses associated with ageing will increase sooner than expected. Thus, subtle memory losses, physical tiredness, general frailty, as well as specific illnesses may be present when a person with Down's syndrome is in his/her thirties rather than his/her sixties.

This idea of premature ageing is also important as studies of both the biology of ageing and of environmental and social influences on ageing may lead to particular strategies or treatments which would help modify this process and may therefore be of benefit to people with Down's syndrome.

Current research in this area seeks not just to improve life expectancy in general, but to improve what is called "disability-free life expectancy". In other words, as life gets longer this should not be associated with an increase in the disabilities associated with later life.

Health in later life

The second explanation as to why people with Down's syndrome do not tend to live as long as others concerns the possibility that they have a higher rate of illnesses that are associated with an increased mortality, and that these illnesses occur earlier in life, than in those without Down's syndrome. There is mixed evidence to support this. First, the commonest causes of death affecting older people in the general population are heart disease (heart attacks), vascular disease (strokes), and the cancers of later life (breast, gut, lung, prostate, etc). There is no

convincing evidence that people with Down's syndrome have an increased or earlier risk of developing such illnesses. Some, such as heart and vascular disease, may even be decreased, possibly because fewer people with Down's syndrome smoke or because their blood pressure tends to be normal. The only illness known to be associated with a decreased life expectancy and which clearly does appear to occur earlier in life in people with Down's syndrome is a form of dementia called Alzheimer's disease. These observations suggest that in people with Down's syndrome the relationship between possible premature ageing and the observed shortened life expectancy is more complex than previously considered.

Alzheimer's disease

In 1907, Alois Alzheimer first described the appearance of brain tissue observed under the microscope from a lady who prior to death had deteriorated significantly in her abilities. The microscopic "plaques" and "tangles" that he observed in the brain tissue are now recognised as the pathological hallmarks of a form of dementia that has come to be known as "Alzheimer's disease". In the past, the loss of memory and skills was frequently referred to as "senility". Alzheimer's disease is now recognised to be the commonest form of a number of disorders that have a similar course, referred to as 'the dementias'. Other causes of dementia include multiple small strokes (referred to as multi-infarct dementia) or (rarely) deficiency of vitamin B12 or brain tumours. Very often someone with Down's syndrome that is showing a deterioration in his/her abilities will not be showing early signs of dementia, but will instead be suffering from something which presents with similar symptoms but are not progressive and can be treated. These conditions include thyroid disorders and depression. For more detailed information, refer to our accompanying leaflet, "Ageing and its Consequences for people with Down's syndrome". It is important to note that a diagnosis of dementia cannot be given until the other possibilities have been ruled out. There can be important differences in the way the different causes of dementia present themselves but the common characteristics of the 'dementias' include : -

CHARACTERISTIC FEATURES OF DEMENTIA

Progressive loss of abilities over time including:

- Memory: most characteristically memory for recent events
- Higher mental functioning: deterioration in the ability to plan and think and to undertake specific complex tasks such as dressing oneself (referred to as dyspraxia)
- Language: increasing inability to understand or use language appropriately (referred to as dysphasia)
- Living skills: loss of ability to undertake previously acquired tasks of day to day functioning such as caring for oneself, making a drink, cooking etc
- Changes in personality: changing moods, irritability, stubbornness etc

Alzheimer's disease is the form of dementia that characteristically has a progressive course and, if brain tissue is examined after death, it is found to have abnormal numbers of microscopic plaques and tangles and the brain size has shrunk considerably. It is this 'atrophy' of brain tissue that leads to the loss of skills.

Not long after Alzheimer reported finding these plaques and tangles in the brain of the lady he studied, who clearly had dementia, similar findings were also reported in the brains of people with Down's syndrome who had died. In the case of those with Down's syndrome these plaques and tangles were found to be present much earlier (in their twenties and thirties) than was the case in those without Down's syndrome.

It is now clear that the risk that a person with Down's syndrome may develop the symptoms of dementia associated with the development in the brain of these 'Alzheimer' plaques and tangles increases with age from the age of about thirty. By their fifties approximately half of the people with Down's syndrome will be showing evidence of memory loss and other problems (such as loss of skills) associated with having Alzheimer's disease. The risk that a person with Down's syndrome will develop Alzheimer's disease increases with age at a similar rate as it does in the general population but 30 to 40 years sooner than is the case for people without Down's syndrome.

When should you suspect the possibility of Alzheimer's disease?

Alzheimer's disease, as with all the dementias, affects a person's ability to think and reason, to remember things and to undertake the tasks necessary for everyday life. It can be difficult to detect its onset and early course; in people with Down's syndrome it may affect their personality first before the more classical early features of memory loss become obviously apparent. This is illustrated with two examples below: -

1. Mary had lived in a group home for people with learning disabilities for the last twenty years, having moved there after her mother died suddenly when Mary was 35. She has led a relatively independent and active life, travelling by herself on the local bus to a day centre and also attending social events in the evening and at weekends. She is now 55 and, mainly over the last six months, those supporting her have become concerned because she has changed. She says she is happy but others have noticed that she has become rather irritable and more 'stubborn' than before. There have been two occasions when she got off the bus at the wrong stop and appeared very bewildered and lost. The staff at her home noticed that, very unusually for her, she forgot her father's and her brother's birthdays this year. She herself has also noticed that her memory is not as good. More recently she came downstairs having put her top on the wrong way round. Although she realised it 'wasn't quite right' she was unable to appreciate precisely what she had done wrong and how it should be corrected. Her physical health was fine and there had been no major changes in her life.

2. Paul is now in his late forties. He has always had significant learning disabilities as a result of having Down's syndrome. He continues to live with his elderly mother with some additional support from other family members and some paid care. He has attended the local day centre for people with learning disabilities for many years. His mother has noticed that he has become more moody and irritable and often now refuses to help with the day-to-day tasks he has enjoyed in the past. At the day centre he was always known as 'a bit of a loner' but he has become

more isolated than usual and increasingly irritable if approached by others, even when he knows them well. He has started to wander into the wrong rooms and was found trying to urinate in an office. He has also wandered out of the day centre along a busy road. These incidents are all very unlike him. Even though he has always needed some help with organising his day it is clear to the staff that he is even less able to do this now and seems to forget what he has to do next. Whilst he has been a bit tearful his mother and the staff do not think he is depressed. He has limited spoken language but with the help of those who know him well he does not report any specific complaints. A month ago he had a sudden episode of confusion lasting a brief period of time. The GP wondered whether this might have been a seizure. Other than this, physically he has been well.

These two examples illustrate the sort of changes that should initiate a clinical assessment. The questions to be answered are as follows: Why have these changes occurred? Does he/she have any specific problems that would be resolved through specific treatment for a condition such as depression or thyroid disorder? If it is likely that he/she is developing Alzheimer's disease, what are the implications for treatment and support?

Obtaining a diagnosis

It is the person with Down's syndrome, his/her family and/or paid carers that will invariably be the people who first observe such change. This should trigger an assessment, initially by the GP and, if necessary, through a referral to a local specialist service. The diagnosis of dementia and specifically of Alzheimer's disease depends on the following:

- Evidence of a pattern of change in personality and general functioning that is characteristic of dementia, such as evidence of a deterioration in memory, general mental functioning, living skills, and personality.

- The exclusion, through a detailed history, clinical examination, and the undertaking of appropriate investigations, of other causes of dementia or of other disorders that might mimic dementia. For more information on some of these other disorders, see our accompanying booklet, "Ageing and its Consequences for People with Down's Syndrome".

Assessments

The diagnosis of dementia and the exclusion of those disorders that might mimic dementia depend upon a detailed description of what has been observed by those that know the person well and also a full medical and psychological assessment of the person him/herself.

Informant interview

People who are developing dementia may be able to describe some of the changes that have occurred to them (such as an awareness of memory loss) but many may not be able to remember sufficient details about exactly what has happened over time. This may particularly be the case for people with learning disability and dementia. Information from someone who has known the person with Down's syndrome, ideally for years but at least for six months, is therefore very important. If the person with Down's syndrome is no longer able to describe for him/herself what he/she was able to do in the past, the observations of someone who knows him/her will be the main source of that information. Sometimes information in life-story books or from other sources can help supplement the observations of others. When determining whether the changes observed in the person with Down's syndrome might be due to dementia or to any other illness the doctor undertaking the assessment will want to know about whether deterioration or change of any sort has been observed in the following areas:

- Memory
- General mental functioning
- Specific abilities and living skills
- Personality
- Mood and general behaviour
- General physical health
- The person's living and family circumstances

Assessment of the person with Down's syndrome

The following broad areas of assessment are required: -

- The person's own description of any changes in their mood and general well-being and physical state
- An assessment of his/her cognitive abilities (such as memory) using an established neuropsychological test developed for assessing those suspected as having developed dementia (e.g., the CAMCOG, the Severe Impairment Battery)
- Physical examination

The second of these (neuropsychological assessment) requires the person to answer questions to test his/her memory, whether he/she is fully orientated in time and place, whether he/she can copy shapes, follow instructions etc. Such assessments can be particularly important repeated over time as these repeat tests can detect the nature and extent of any deterioration in the person's cognitive abilities such as memory, use of language or orientation.

Investigations

The exact investigations arranged will be guided by the results of the above assessments but may include:

- A blood test to check thyroid function, to rule out conditions such as anaemia or problems with kidney or liver function, or vitamin B12 deficiency (that in rare cases can cause a dementia-like illness);
- Vision and hearing assessments if sensory impairments are suspected;
- MRI or CT brain scan if there is uncertainty about whether the changes are due to dementia, or if the emerging picture is unusual, or suggests the very rare possibility of some other brain problem that might explain the observed changes (e.g., brain tumour).

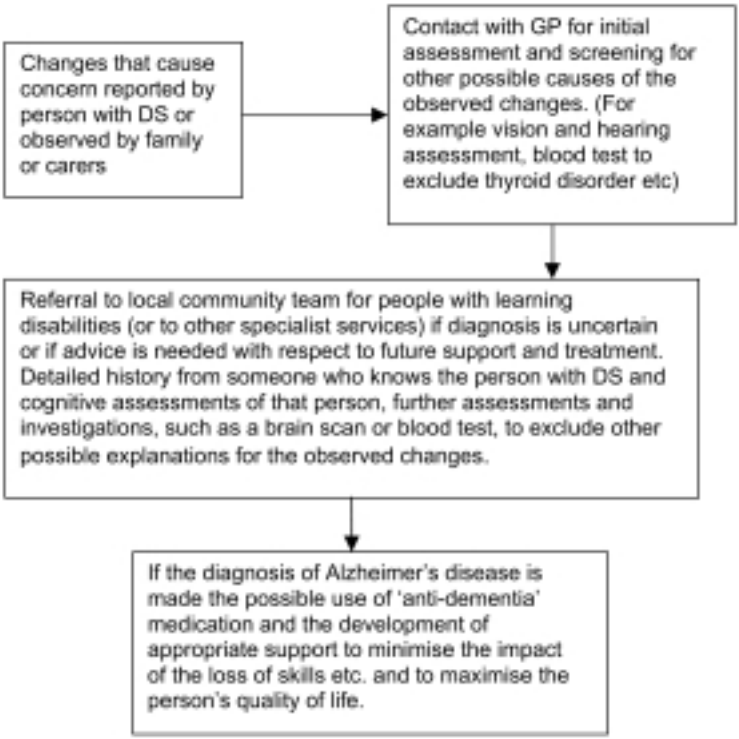
Given the types of changes described earlier, these are the main problems that should be considered. This is referred to as the differential diagnosis.

'Differential diagnosis' of behavioural and functional change in later life in people with Down's syndrome

- Depression
- Under-active thyroid gland (hypothyroidism)
- Sensory impairments (visual and hearing loss)
- Dementia (usually Alzheimer's disease)
- Impact of major life events such as bereavement
- Other rare illnesses

The process that ultimately leads to a diagnosis of Alzheimer's disease and what happens next is illustrated in Figure 1.

Figure 1



Course of Alzheimer's disease affecting people with Down's syndrome

Alzheimer's disease is a progressive brain disorder that results in the loss of normal brain structure and function progressively over time. For this reason the symptoms of Alzheimer's disease change as the illness progresses. In the general population the progression of Alzheimer's disease has generally been divided into three parts. In the case of people with Down's syndrome the early presentation may be different from those with Alzheimer's disease in the general population but the later course appears very similar. These stages are summarised below: -

Course of Alzheimer's disease in people with Down's syndrome

Stage 1
Changes in behaviour and personality, such as unexplained and uncharacteristic stubbornness, changeable moods, oddities in behaviour, together with subtle evidence of deterioration in memory for recent events and deterioration in general mental functioning.

Stage 2
Obvious deterioration in memory and evidence for a significant deterioration in general mental functioning, specifically in the areas of ability and communication. Inability to spontaneously recognise familiar people. Onset of problems such as incontinence and evidence of obvious disorientation and confusion.

Stage 3
Complete loss of self-care, living skills, and ability to communicate effectively, leading to the need for 24-hour care and help with all basic aspects of day-to-day life such as feeding, use of the toilet, hygiene etc. Inability to recognise familiar people or surroundings.

Treatment and strategies for support

As yet there are no treatments that reverse or significantly arrest the plaque and tangle formation and the brain atrophy that cause the symptoms of Alzheimer's disease. Treatment is therefore aimed at slowing down the progression of the brain deterioration, minimising the impact of the increasing impairments, and maximising the quality of life of the person during the course of his/her illness.

Specific dementia treatments and support strategies

There are now medications that can bring temporary benefits in terms of some improvement in functioning and slowing of the deterioration. Donezepil is one that has been tried in people with Down's syndrome who have dementia and has been shown through research to have some beneficial effects. This class of medication helps to increase one of the brain chemicals (acetylcholine) that is depleted in those with Alzheimer's disease. Newer 'anti-dementia' medications for those with more advanced illness have other types of actions (e.g. memantine). These have not been specifically assessed in people with Down's syndrome. In the longer term it is hoped that there will be treatments that can be safely given and which prevent the Alzheimer's disease developing in people with Down's syndrome or are more effective at slowing its course. As with all medications there is always a balance to be struck between the potential for side effects and the likelihood of benefit. Careful use of medications, usually starting at lower than normal doses, can be justified.

The key to effective support of a person with Down's syndrome who has Alzheimer's is understanding both the person and the disease enough to be able to anticipate his/her difficulties. For example, if the person's memory is deteriorating then this might be compensated for in various ways, such as sensitively and regularly reminding the person who you are and where he/she is. Gesture, signing, and pictures, in addition to using spoken language, may improve the person's understanding and communication thereby helping him/her participate and in turn reduce any anxiety he/she might experience because of his/her confusion and inability to understand.

General physical and mental health

Dementia usually develops at a time in a person's life when he/she is also at risk of other physical and mental disorders. The occurrence of such disorders will compound any difficulties that are a consequence of the person's dementia. In addition, the presence of dementia itself (particularly in the advanced stages) may result in poor physical health due to poor fluid and food intake, or the risk of urinary and chest infections. Regular reviews of both physical and mental health are therefore essential. Keeping as healthy as possible helps to minimise the impact of dementia. Infections (such as urinary or chest infections) can

cause the person in the early stages of dementia to get even more confused. A sudden deterioration in the person's general mental state should therefore raise the possibility that they have acquired a physical illness. Similarly, the use of certain medications (such as sedatives or for the treatment, for example, of heart problems) may be associated with increased states of confusion when a person has the additional problem of dementia. Thus, all medication must be used with caution.

Changes to the living environment

People with dementia have increasing difficulties remembering new information. What happened yesterday or first thing that morning, who people are or where he/she lives, may all be a mystery to the person with dementia. It is memory that gives structure and meaning to our lives and without it, or if it is patchy, the world becomes confusing and frightening. For this reason, in general, people with dementia are best supported in places where they have lived for some time. This of course may not always be possible.

The environment is not only important because of the person's deteriorating memory but also because of other problems that arise. Confusion is often worse at night because people cannot use visual clues to help orientate themselves - so lighting is important. The type of carpet, whether doors to rooms are easily distinguished from one another and whether electrical and other appliances are safe are all important questions. These issues are summarised below: -

The environment

- Are there any physical or other hazards in the kitchen, bedroom or other rooms that require changes to be made?
- Are there changes that can help the individual to remain orientated in the living space and recognise his/her room, the toilet etc?
- Can lighting and safety be improved at night-time?
- Are the living and other relevant environments reviewed regularly in the light of the person's changing needs?

Additional sources of support

As the needs of the person you care for change and the level of care he or she requires increases, it may be useful to have a Community Care Assessment carried out by Social Services. Social Services will then work closely with health services and other organisations where necessary to ensure that the appropriate level of care is given. You can also request a Carer's Assessment to look into your own needs. For more information on these contact your local Social Services, or call the DSA National Office on 0845 230 0372.

For further information on the behaviour of someone with Down's syndrome and dementia, and for detailed information on how to support and care for that person, we recommend the Down's Syndrome and Dementia Resource Pack published by the British Institute of Learning Disabilities and written by Karen Dodd, Vicky Turk and Michelle Christmas. ISBN number 1-90408-237-8; call BILD on 01562 723010 for more details.

Further Information

The Foundation for People with Learning Disabilities
83 Victoria Street
London SW1H 0HW
Tel. - **020 7802 0300**
www.learningdisabilities.org.uk

British Institute of Learning Disabilities
Campion House
Green Street
Kidderminster
Worcestershire DY10 1JL
Tel. - **01562 723010**
www.bild.org.uk

Mencap
123 Golden Lane
London EC1Y 0RT
Tel. - **0808 808 1111**
www.mencap.org.uk

Disabled Living Foundation
380 – 384 Harrow Road
London W9 2HU
Tel. – **020 7289 6111**
www.dlf.org.uk

Alzheimer's Disease Society
Gordon House
10 Greencoat Place
London SW1P 1PH
Tel. – **020 7306 0606**
www.alzheimers.org.uk

Dementia Services Development Centre
University of Stirling
Stirling FK9 9LA
Tel. – **01786 467740**
www.stir.ac.uk/esdc

Housing Options
78a High Street
Witney
Oxfordshire
OX28 6HL
Tel. – **0845 456 1497**
www.housingoptions.org.uk

Mind
Granta House
15 – 19 Broadway
Stratford
London E15 4BQ
Tel. – **0845 766 0163**
www.mind.org.uk

Cruse Bereavement Care
126 Sheen Road
Richmond
Surrey RW9 1UR
Tel. – **020 8940 4818**
www.crusebereavmentcare.org.uk

Down's Syndrome Scotland
158 – 160 Balgreen Road
Edinburgh
EH11 3AU
Tel. – **0131 313 4225**
www.dsscotland.org.uk

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Tony is a consultant Psychiatrist specialising in seeing people with learning disabilities. He has been the psychiatric adviser to the DSA for many years. He trained in medicine at University College London, qualifying in 1973. After some years in general medicine he trained in psychiatry at the Maudsley Hospital and Institute of Psychiatry. He first undertook research on Down's syndrome and ageing at that time. He moved to the University of Cambridge in 1992 and since 2002 he has been the first holder of the PPP Foundation Chair in Learning Disabilities.

HELP FOR PEOPLE WITH DOWN'S SYNDROME

DOWN'S SYNDROME ASSOCIATION
A Registered Charity

If you would like further information on the Down's Syndrome Association, or are interested in joining as a member, please give us a call on the number below.

National Office:
Langdon Down Centre,
2A Langdon Park, Teddington, Middlesex TW11 9PS
Tel: 0845 230 0372
Fax: 0845 230 0373
Email: info@downs-syndrome.org.uk
Website: www.downs-syndrome.org.uk

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Most of our literature is available free of charge on www.downs-syndrome.org.uk. However, each piece costs us **£2.90 t**o write, produce and post in printed form.

If you can, **please make a donation** of any amount so we can continue to supply information free to those who need it. Thank you.

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