Down's Syndrome and Alzheimer's Disease.

A Multidisciplinary Pack to raise awareness and promote efficient diagnosis.

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Life Expectancy

Advances in medical, surgical, nursing and social care in recent years has resulted an ever-increasing life expectancy, and an aging population. This has given rise to individuals with Down's syndrome developing functional decline in what the general population calls middle age. While initial observation of this decline was originally reported over one hundred and thirty years ago, it was not until relatively recently that a link between DS and Alzheimer's disease was widely recognised.

Many authors (Fryers, 1986, Demissie et al, 1988, Carr, 1994, Bar and Campbell, 1995) have commented on the life expectancy of individuals with Down's syndrome. Hutchinson (1999) suggests in 1929 the average was just 9 years old, while Jancar (1988) suggests in 1945 the average was between 20 and 30 years of age. Demissie et al (1988) state at the time of their work the oldest person with Down's syndroem was 75 years old, indeed Barr and Campbell (1995) advise the average life expectancy today is around 60-70 years old. Carr (1994) and Jancar (1988) suggest this is due to the advances in medical, surgical and nursing care available, along with the increased respect for individuals rights (Wolfensburger, 1972).

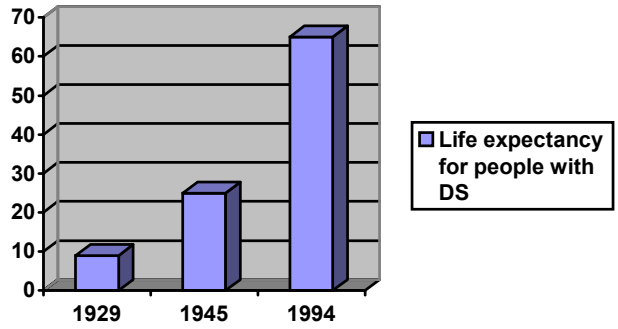


Figure 1 Graph showing Life Expectancy of individuals with DS (Hutchinson, 1999; Jancar, 1988; Barr & Campbell, 1995)

However Holland (1998) recognizes the mean life expectancy is still lower than that of the general population, which according to South East Public Health Office (2002) is currently 76 years of age.

Due to increasing life expectancy, several authors (Steffelaar & Evenhuis, 1989, Prasher 1995, Campbell and Barr 1995) have commented on the population increase of individuals with Down's syndrome, indeed Janicki and Dalton (1998) point out that currently the percentage of older people in developed nations is between 12-20%, which Kinsella and Tauber (1993) advise will rise to around 30% in the next 20 years. Evans (1990) advises these statistics pose a significant public health policy challenge. Hutchinson (1999) concurs relating the increasing life expectancy of individuals with Down's syndrome to the apparent increased risk of Alzheimer's Disease. This association is not new, Fraser & Mitchell (1876, cited in Mann, 1994) initially observed the signs of a "precipitated senility" (Oliver & Holland, 1986, p307). Jervis (1948) provided the first major study, concluding from autopsy that three individuals with Downs's syndrome, who had exhibited this functional decline, had characteristics of Alzheimer's disease as early as 30 years of age. However, Barr & Campbell (1995) observe these findings were not widely distributed, indeed, with reference to the life expectancy graph - it could also be suggested individuals with Down's syndrome may not have been reaching the age of this functional decline consistent with Alzheimer's disease.

The Link between Down's syndrome and Alzheimer's disease

The link between Down's syndrome and Alzheimer's disease remains unclear, however many authors (Ball & Nuttall, 1980; Lai & Williams, 1989; Mann, 1994; Holland, 1994; Percy, 1998) have commented upon the similarity of the neuropathological changes between individuals with and without Down's syndrome. This work has centered on the association areas of the frontal, temporal and parietal areas of the brain. Percy (1998) also states that all individuals with Alzheimer's disease display an accumulation of beta amyloid protein around the blood vessels in the brain, interestingly this protein is encoded by a gene on chromosome 21 (Holland, 1994). This led Mann (1994) to speculate in the Down's syndrome population, the extra chromosome 21 caused the excess production of the beta amyloid protein and the subsequent development of Alzheimer's disease type symptoms. While this seems to link the two, Holland (1994) reserves judgment suggesting it is unclear as to whether the deposition of the protein is a cause or a result of Alzheimer's disease. Indeed Percy (1998) discusses the several other predisposing factors to Alzheimer's disease, including head injury, the female sex, depression, hypothyroidism, electromagnetic radiation, poor nutrition, exposure to toxic metals, other brain poisoning chemicals such as alcohol, anti-convulsant medications, solvents and herbicides.

Implications for Practice

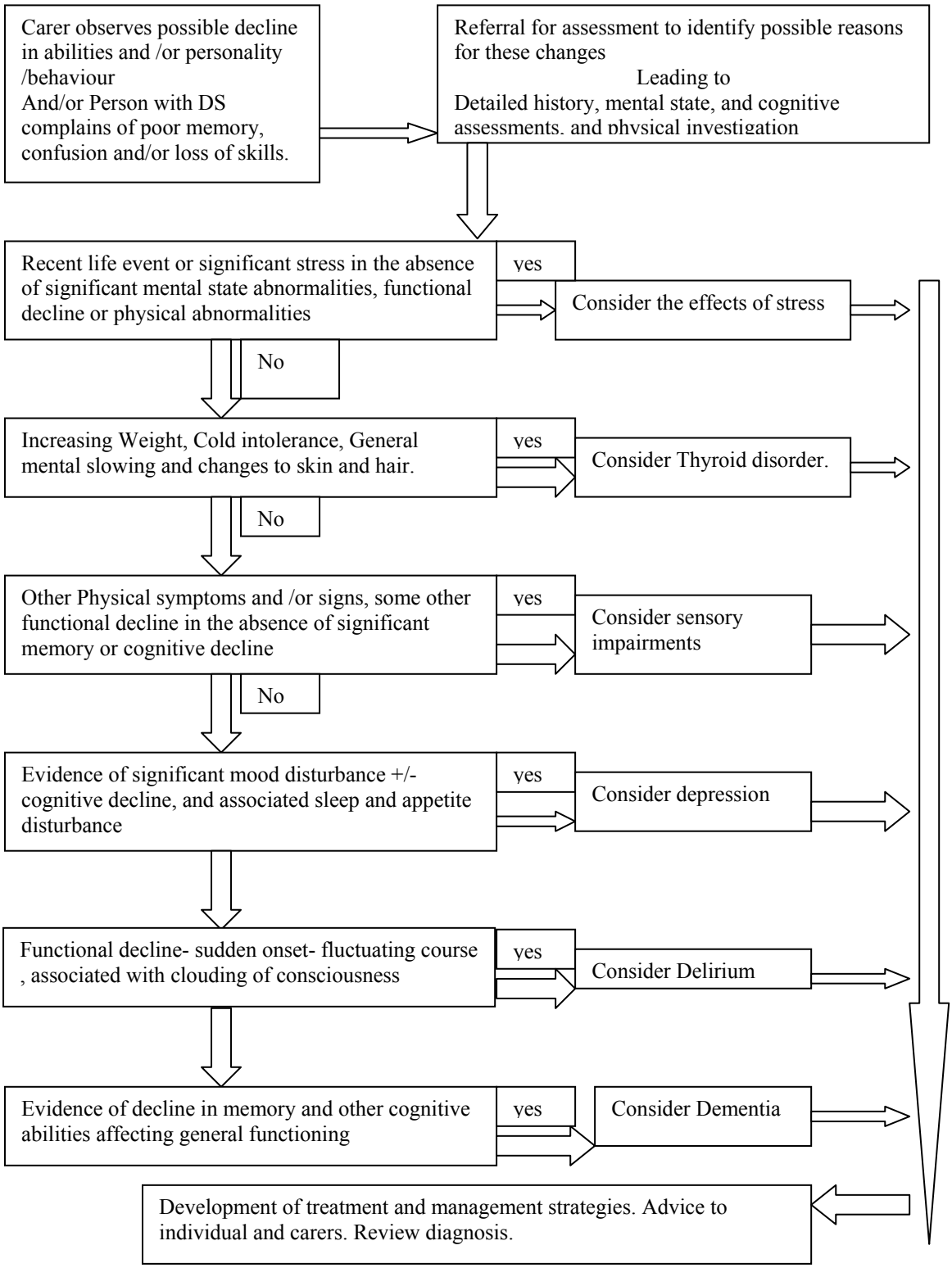
As a Community Nurse, the author is aware of the increasing number of referrals to the Community Team for People with Learning Disabilities (CTPLD) in regard to functional decline. These cases often require collaborative work (Paxton, 2001) with individuals within the CTPLD and external professionals such as General Practitioner, the local Mental Health Team, and the Neurology Department within secondary care. Kerr's (1997) and Lishman's (1998) observe that the clinical features of Alzheimer's disease initially start with a gradual onset with a slow progressive decline in functioning, make it difficult for carers to recognise any differences until Alzheimer's disease has taken complete control. This can cause difficulty with diagnosis, and certainly a great deal of distress for the client and carers. Indeed Holland (1994) notes that, although the neuropathological changes may be evident when an individual reaches the age of 35, the clinical signs do not appear until much later. It could be suggested that the clinical signs of Alzheimer's disease may well not be recognised in individuals with Down's syndrome until the more obvious symptoms of functional decline have taken hold. Holland (1998) recognises the effective diagnosis is critical for appropriate support strategies to be put in place. As a community nurse, the author may well be involved in this process, however there are several health issues need consideration as they may present similar to those of Alzheimer's disease.

Paradoxically, now a link is recognised between Alzheimer's disease and Down's syndrome, diagnosis should not be presumed, as an apparent decline in abilities could be caused by several other health issues that could be more successfully treated. Many authors (Holland, 1995; Holland, 1998; Barr & Campbell, 1995; Donnelly & Earnshaw, 2002; McKenzie, 2000; Paxton, 2001) have noted the need to screen for hypothyroidism, as this can cause similar symptoms, and occurs in as many as 30% of individuals with Down's syndrome (Holland, 1995). Similarly, depression can also cause symptoms akin to Alzheimer's disease, indeed Burt (1998) advises that depression can co-exist with a dementia. desRosiers (1992a, 1992b) suggests even in the general population primary dementia and dementia secondary to depression are mistaken for each other at least 10%-20% of the time, Burt (1998) concurs advising this risk of misdiagnosis increases when applied to individuals who

have an intellectual disability. Indeed Holland (1998) identifies several issues that could cause apparent Alzheimer's disease symptoms, and provides an assessment pathway for age related changes in people with Down's syndrome.

Holland (1995) advises that although an individual with Down's syndrome may have a significant intellectual impairment, they will have a range of abilities, but the decline in these may go unrecognized due to the difficulty differentiating these from the learning disability. This is particularly relevant to the author's practice as individuals referred for assessment for cognitive decline are often unknown to the nurse involved, causing assessment to be based on carers reports. This information can vary in quality due to high staff turnover in residential services (McKenzie, 2000).

Assessment of Age Related changes in people with Down's Syndrome



Diagnosis of Alzheimer's disease

Several assessment tools available can provide varied information regarding an individual's skills, emotional changes, and functional decline. The author would recommend that these could be categorised by time scale, and urgency, as well as training to use such tools.

The first stage of assessment needs to become part of person centred planning (DoH, 2002a), and health action planning (DoH, 2002b), that intimate that individuals with learning disabilities should have the same rights in the National Health Service and therefore have the same responsibilities for their own health. Several tools could be used at this point by staff who have minimal training with service users. The most effective of which -the author believes to be- the STAR profile.

The Social Training Achievement Record (STAR) Profile (Williams, 1986).

This assessment is easy to use with some training, and although is not time efficient, results can be displayed graphically for ease of comparison, in longitudinal studies. The assessment is based upon activities of daily living, and is criterion based for baseline and re-assessment scores of functional strengths and needs.

Carers -both residential and family- could complete this assessment, with people with Down's syndrome, several years before the individuals fortieth birthday, providing a baseline, and a regular review of skills. Alzheimer's disease initial symptoms are often difficult to observe, as they are quite slow moving, and regular reviewing using an assessment of this kind can provide carers with evidence and confidence to approach primary care and specialist services.

The PAS-ADD (Hester Adrian Research, 1992)

PAS-ADD stands for Psychiatric Assessment Schedule for Adults with Developmental Disability. Use these three schedules to improve the detection and diagnosis of mental health problems in people with learning disabilities. The multi-level system consists of three components:

(a) The Psychiatric Assessment Schedule for Adults with a Developmental Disability (PAS-ADD). This is a semi-structured clinical interview for use with respondents

who have mental retardation and for key informants. It uses a computer algorithm to produce diagnoses and other diagnostic information under ICD 10, this is for use by Psychiatrists and psychologists.

(b) The PAS-ADD Checklist. This is a psychiatric symptom checklist for use by direct care staff and families. Similar to the STAR profile, this checklist provides support with identifying psychiatric symptoms and evidence with which to show the GP or other health professional.

(c) The Mini PAS-ADD. This is for use by staff who do not need to have a professional background in psychopathology, but should have received some training in its use. With training nurses and allied health professionals can use the Mini PAS-ADD assessment tool. The use of this tool should be able to identify issues of functional decline.

Dementia Questionnaire for Persons with Mental Retardation (Evemhuis, 1995)

This assessment is similar to the STAR profile as it is most effective as a screening tool. However, the subjects covered are more applicable to screening for functional decline and has been studied specifically for sensitivity. A health professional and carers in partnership most effectively complete the questionnaire. It is possible for a psychologist or psychiatrist to diagnose dementia on one use of this tool however it would be in comparison of previous Intelligence Quotient assessments. This would be an ideal tool for a specialist team screening program, as it is designed especially for screening for dementia in people with learning disabilities, is less time consuming than some of the more in depth packages available.

Adaptive Behaviour Scale – Residential and Community (Nihira, Leland & Lambert, 1993)

This assessment has been used in a number of academic studies examining the relationship between Down's syndrome and Alzheimer's disease (Aylward et al, 1995; Burt et al, 1992; Cooper, 1997; Crayton et al, 1998; Hon et al, 1998; MCkenzie et al, 2002) and has been recommended as an assessment tool in the good practice guidelines developed by Aylward et al (1995). However, it is time consuming-assessing 297 items, and is recommended to be carried out by a psychologist, which

may not always be an efficient use of locality resources. Again, it is recommended this assessment be used longitudinally for effective diagnosis, however it does provide information pertaining to the functional and behavioural characteristics of the individual's decline. McKenzie et al (2002) also recommend an alternative scoring system for this assessment (see appendix 1) that provides more sensitive results; completed at regular intervals this method will provide a more detailed picture of an individuals loss of skills, and hence provide more evidence for diagnosis.

The Mini Mental State Examination (Folstein wt al, 1973)

This assessment is used in the general population for diagnosing dementia, however it is not always appropriate for use with individuals with learning disabilities, as it presumes the individual was of a 'normal' cognitive ability prior to the decline. Indeed the questions assume an ability to read, write, and to subtract numbers methodically, which may not be applicable for some individuals we work with. It should also be noted at this point the National Institute for Clinical Excellence Guidelines on the Use of Donepezil, Rivastigmine and Galantamine (2001) base the prescription of these anti-Alzheimer medications on the use of this assessment. As such this could be interpreted a discriminatory.

Short NeuroPsychiatric Inventory Booklet (Cummings et al, 1994)

This is a twelve-stage assessment tool that focuses on the psychiatric and behavioural characteristics of an individual. This tool -like the PAS-ADD- may support differentiation between mental health problems i.e. depression and dementia. However, this tool is unable to detect functional declines, and as such may well need to be used in conjunction with a skills assessment. It is recommended that this assessment is used by a psychiatrist, however the questions and prompts are self explanatory and as such could be used by health professionals generally.

It should also be noted that Aylward's (1995) good practice guidance also advises on the use of several other assessment packages of which the author has been unable to obtain, these include:-

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