
Down Syndrome and Alzheimer's Disease: Dual Diagnoses

Alzheimer's disease is a progressive, degenerative brain disorder, which affects individuals' abilities to think, reason, remember, use language and know where they are in space and time. Eventually, affected persons are unable to recognize family, become bed-bound, mute, incontinent, and ultimately require total care.

The course of the disease can last 3-20 years, affecting all areas of family life. It is a disease that imposes serious financial strains on the family and society, and yet there is little assistance from any insurance carriers, including Medicare, to help families care for their family members at home. Most of the cost of care is borne by families, individuals, or the Medicaid system.

The cause of Alzheimer's disease is still unknown, and to date, the treatment is medication for symptom management, as well as education and support throughout the course of the illness for individuals and family members affected by Alzheimer's disease.

Approximately 4 million Americans are affected by Alzheimer's disease, including 10% of adults age 65 and older and approximately 40% of individuals over the age of 85. Alzheimer's disease affects both men and women, but women particularly are more vulnerable.

The known risk factors for Alzheimer's disease are:

- ◆ Age
- ◆ Family History
- ◆ Genotype
- ◆ Down Syndrome

The Down Syndrome Connection

Down syndrome appears in 1 to 733 live births and accounts for approximately 15% of cases of mental retardation. As recently as the 1930's, the average life expectancy of persons with Down syndrome did not extend beyond the early 20's. Despite the accelerated aging process that occurs in persons with Down syndrome, advances in medical, technological and personal care procedures have contributed to approximately 70% of individuals now living beyond their 50th birthday.

Nevertheless, it is known that individuals over 40, who have Down syndrome and who have a family member with Alzheimer's disease, are at greater risk of developing Alzheimer's disease. In contrast, most adults with mental retardation are at the same risk for developing Alzheimer's disease, as are individuals in the general population.

Shared Susceptibility

The neuropathological hallmark lesions (tangles and plaques) of Alzheimer's disease are present in the brains of all adults with Down syndrome by the age of 40 years, which suggests a shared genetic susceptibility to Down syndrome and Alzheimer's disease. However, these characteristic tangles and plaques do not necessarily mean that all individuals with Down syndrome will develop Alzheimer's disease. Nonetheless, it is well documented that the prevalence of dementia increases with age in persons with Down syndrome. Depending on the particular study that one reads, it is estimated that clinical and behavioral symptoms of Alzheimer's disease present in approximately 50-70% of individuals with Down syndrome by the time they reach 60 years of age.

Clinical Features

The symptoms and progression of Alzheimer's disease in the Down syndrome population present somewhat differently than in the general population. The progression of Alzheimer's disease for persons with Down syndrome takes, on average, about eight years. The symptoms are sometimes subtle and because of the pre-existing cognitive impairments, families and care providers may not notice that something is amiss.

Some of the symptoms family members or care providers may see in a person with Down syndrome are:

- ◆ Personality changes, irritability or apathy
- ◆ Loss of speech or change in language skills
- ◆ Disorientation to time, place
- ◆ Decline in self care skills; this is important because it reflects a person's level of functioning and it is here that a care provider or family can observe over time, a subtle or sudden decline
- ◆ Abrupt onset of seizure activity when there had been none in the past
- ◆ Incontinence, when an individual has always been independent in toileting
- ◆ Short- term memory loss may depend upon the previous level of memory demands and reliance on memory in everyday life
- ◆ Sleep/wake cycle disruptions

Just as in the general population, the course and symptom presentation is unpredictable and unique to the individual.

Diagnosis

The same diagnostic process that applies to individuals in the general population is applicable to those with Down syndrome. However, when there is pre-existing cognitive impairment and variation in a person's functional ability, the diagnosis of Alzheimer's disease is difficult and must be determined thoughtfully and thoroughly.

A determination of probable or possible Alzheimer's disease can be reached through a focused, comprehensive historical, medical, neurological, and neuropsychological assessment.

The clinician must:

- ◆ Rule out and treat all possible causes for the observed changes; for example, hypothyroidism, sensory losses, acute medical condition (e.g. urinary tract infection), constipation or over medication.
- ◆ Diagnose and treat depression/dementia syndrome.
- ◆ Be aware that common neurocognitive diagnostic tests used in the general population are not useful in individuals with Down syndrome or mental retardation.

Clinicians must consider all of the above, treat that which is reversible, and then conduct periodic assessments over time.

Importance of Obtaining a Baseline Assessment

Ideally, periodic screenings with identification of baseline skills and functional status should be conducted regularly (yearly after age 40) so that, should changes occur, the baseline measures and life histories will highlight the significance of those changes or losses.

Treatment and Supportive Care

As with Alzheimer's disease in the general population, there is no cure, however supportive treatment may include cholinesterase inhibitors (e.g. Aricept or Exelon), medications for the psychiatric and behavioral symptom management, and ongoing education and support to family and care providers. Often, the person with Down syndrome is living with an elderly parent or sibling. These family members have advocated and cared for their children/siblings all of their lives and are now faced with multiple issues and concerns, while they have little energy and limited resources to deal with them.

It is critical that all families and group home providers develop a care plan that is realistic, appropriate, and will meet the needs of both the individual and family/care providers.

As in the general Alzheimer's population, this involves:

- ◆ planning for the legal and financial future of the individual
- ◆ planning for the future long-term supportive housing/care needs of the individual so that s/he has the opportunity to "age in place"
- ◆ addressing the delicate balance that exists between preserving a person's autonomy, yet providing the supports necessary for maintaining the person in a safe emotional and physical environment
- ◆ learning how to care for the individual throughout the course of the disease and,
- ◆ recognizing and addressing the respite needs of all formal and informal caregivers

What can the Greater Cincinnati Chapter of the Alzheimer's Association do for Affected Individuals and Family/Care Providers?

The Greater Cincinnati Chapter of the Alzheimer's Association provides both educational and supportive programs to providers and families who are caring for and working with individuals with Down syndrome and probable Alzheimer's disease.

Families and providers can meet with a clinical nurse specialist to discuss:

- ◆ concerns over changes in behavior, personality or self-care skills seen in the individual with Down syndrome or developmental disability
- ◆ importance of establishing baseline assessment and subsequent differential diagnosis
- ◆ educational and supportive needs of family or provider
- ◆ family-directed, individualized care planning which enables the provider/family to provide appropriate care in the present and throughout the course of the disease process

If you are interested in speaking with someone about your particular situation, please call Clarissa Rentz MSN, APRN, at the Greater Cincinnati Chapter at (513) 721-4284.

RESOURCES

Down Syndrome Association of Greater Cincinnati

644 Linn Street
Cincinnati, Ohio 45203
(513) 761-5400

American Association on Mental Retardation

ATTN: Alzheimer Disease Workgroup
444 North Capitol Street, NW Suite 846
Washington, DC 20001-1512
1-202-387-1968
www.aamr.org/index.shtml

National Down Syndrome Society

666 Broadway
New York, New York 10012
Helpline: 1-800-221-4602
1-212-460-9330

The Arc

National Headquarters
1010 Wayne Ave. Suite 650
Silver Spring, MD 20910
301-565-3842
301-565-5342
info@thearc.org

REFERENCES

Reports of the AAMR-IASSID Workgroup

"Diagnosis of Dementia in Individuals with Intellectual Disability"

"Practice Guidelines for the Clinical Assessment and Care Management of Alzheimer and other Dementias among Adults with Mental Retardation"

"Epidemiology of Alzheimer Disease in Mental Retardation: Results and Recommendations from an International Conference

Copies may be obtained by contacting the:

The American Association on Mental Retardation

ATTN: Alzheimer Disease Workgroup

444 North Capitol Street, Suite 846

Washington, DC 20001-1512

1-202-387-1968 or

<http://161.58.153.187/Bookstore/Downloadables/index.shtml>

Other References used in this Information Sheet

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Barcikowska, M., Silverman, W., Zigman, W., Kozlowki, P., Kujawa, M., Rudelli, R., & Wisniewski, H.M.(1999). Alzheimer-type neuropathology and clinical symptoms of dementia in mentally retarded people without Down syndrome. Journal on Mental Retardation, 95(5), 551-557.

Janicki, M.P., & Dalton, A.J. (2000). Prevalence of dementia and impact on Intellectual disability services. Mental Retardation, 38(3), 276-288.

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Visser, F.E., Aldenkamp, A.P., van Huffelen, A.C., Kuilman, M., Overweg, J., & van Wijk, J. (1997). Propsective study of the prevalence of Alzheimer-type dementia in institutionalized individuals with Down syndrome. American Journal on Mental Retardation, 101(4), 400-412.

Wisniewski, K.E., Wisniewski, H.M., & Wen, G.Y.(1985). Occurrence of neuropathological changes and dementia of Alzheimer's disease in Down's syndrome. Annals of Neurology, 17, 278-282.

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