

Down Syndrome News, Vol. 27, No. 5

Is it Alzheimer's disease?

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Editor's note: Thanks to Dr. Chicoine, medical director at the Adult Down Syndrome Clinic at Lutheran General Hospital, for writing this article for Down Syndrome News.

"My 24 year old son was diagnosed last week with Alzheimer's disease, what do you think? This is an example of a phone call, an e-mail, or a concern voiced at an office visit that we have been hearing more frequently. Is it Alzheimer's disease? Do people with Down syndrome develop Alzheimer's disease at that age?"

Before answering those questions, it is helpful to go back and look at the information we know about Alzheimer's disease in people with DS.

Alzheimer's disease is a progressive neurological condition. Brain cells are destroyed and the person experiences decline in function in multiple areas including memory, cognition, control of bodily functions and others. Microscopically, the brain tissue demonstrates changes called plaques and tangles.

Several years ago researchers did autopsy studies on a number of people with DS that had died for a variety of reasons. They concluded that essentially all people with DS develop plaques and tangles by the age of 35 or 40. Many people have concluded from this information that all people with DS over the age of 35 or 40 years develop clinical Alzheimer's disease.

We have looked at the information we have gathered after serving over 2,500 adults with DS and have not found that all people with DS over the age of 35 or 40 develop Alzheimer's disease. Some other researchers have published similar findings. In fact, the percentage of our patients who have developed Alzheimer's disease is actually pretty similar to the general population. The difference in our data is that our patients, on average, develop Alzheimer's disease 20 years earlier than people in the general population. Therefore, the rate of Alzheimer's disease in our patients in their forties is similar to those without Down syndrome in their sixties, our patients in their fifties compare to others in their seventies, and sixties to eighties. This is not to say that these are small numbers. In the general population, the incidence of Alzheimer's disease in people in their eighties is thought to be 40 percent.

While this information regarding populations of people is helpful, it doesn't answer the question of whether an individual has Alzheimer's disease. A thorough evaluation is needed. There is no one test that makes the diagnosis. The diagnosis is made by looking

for a pattern of decline, looking for supporting information (such as findings on a CT scan), and “ruling-out” other causes of decline.

There are many reasons someone may have a decline in skills. Some of the causes are not reversible. Unfortunately, as of 2004, Alzheimer’s disease is one of the non-reversible causes. However, many of the other causes are reversible and the evaluation must include an assessment for these. The evaluation includes assessment for such things as:

- Sleep apnea
- Hypothyroidism
- Vitamin B12 deficiency
- Depression

There are many other possibilities as well.

Particularly in younger people who have declined, one of the things we have seen is a person becoming overwhelmed. This is often a case of expectations exceeding ability. We have seen several people who had fine skills in self-care, tasks in the home, and tasks in the work place. Unfortunately, some of these folks have a difficult time organizing their time or dealing with fluctuations in their schedules.

Others have a difficult time knowing how to use their “downtime” or recreation time. This occurs even when the person has the skill to do all the activities but may lack the ability to “pull it together.” This is where the expectations exceed the actual ability. Before the decline, the person appeared to function so well because of the ability to do so many tasks but not as independently as was expected. When the expectations are too great, in a sense, the person “shuts down.”

In Alzheimer’s disease, the symptoms we typically see include the following:

- Memory impairment
- Decline in cognitive skills
- Incontinence of urine and/or stool
- Gait disturbance
- Personality or psychological changes
 - depressed mood
 - aggressiveness
 - paranoia
 - compulsiveness
 - loss of interest in activities
- Seizures and/or myoclonic jerks
- Swallowing dysfunction
- Sleep changes (day-night reversal, day time fatigue)
- Altered appetite and thirst

These symptoms develop over time and not all of them are present in the early stages.

Does the 24-year-old young man mentioned at the beginning of the article have Alzheimer's disease? It certainly would not be the first thing I would consider. In fact, the youngest person we diagnosed with DS was about 35 years old when his symptoms began. Alzheimer's disease is lower on our list of diagnostic possibilities for any of our younger patients who present with a decline in skills. While there may be some people who fall outside the typical age range, it is more likely that there is an alternative explanation. Careful assessment, support, observation over time, and treatment of any potentially reversible conditions will often help make the diagnosis more clear and lead to improvement in those individuals who don't have Alzheimer's disease.