Could it be Alzheimer Disease?

by Adult Down Syndrome Clinic on Monday, October 11, 2010

"My 23 year old daughter with Down syndrome was diagnosed with Alzheimer Disease last week. What do you think?"

This is an example of an email, phone call, or concern voiced at an office visit. Do people with Down syndrome develop Alzheimer Disease at such a young age?

Let’s start with reviewing some information about Alzheimer Disease (AD) and Down syndrome (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 demonstrates changes called plaques and tangles.

Several years ago, researchers did autopsy studies on a number of people with Down syndrome that had died from a variety of causes. 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 develop clinical Alzheimer Disease.

We looked at the data we have gathered after serving about 4500 adults with DS. We have not found that all our patients over age 35 or 40 develop symptoms or signs of clinical Alzheimer Disease. Some other researchers have published similar findings.

The percentage of our patients who have developed AD is pretty similar to those without DS. The difference is that on average, our patients develop AD 20 years earlier. In our practice, the incidence of AD in our patients in their 40’s is similar to people without DS in their 60's (and 50's compared to 70's, and 60's compared to 80's).
While information regarding populations of people is helpful, it doesn't answer the question about whether an individual has AD. 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 data (e.g. atrophy on a CT scan of the brain), 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, at this time, AD is one of the non-reversible causes of decline. However, many of the other causes are potentially reversible and the evaluation must include an assessment for these. Some of these causes include:

- sleep apnea
- vitamin B12 deficiency
- Vitamin D deficiency
- hypothyroidism
- depression

There are other possibilities as well.

Particularly in younger people who have declined, one of the things we have seen is decline due to a person being overwhelmed. This is often the 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 workplace. Unfortunately, some of these individuals have a difficult time organizing their time or dealing with fluctuations in their schedule.

Others have a difficult time knowing how to use their "downtime" or recreational 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, the person may become overwhelmed and "shut down".

There are other reasons why a young person may decline. The members of the Down Syndrome Medical Interest Group are discussing a small subset of these individuals that decline for reasons that are not clear. Further investigation has been initiated to try to understand the cause of decline in these individuals.
In Alzheimer Disease, the symptoms we typically see include:

- memory impairment
- decline in cognitive skills
- incontinence of urine and/or stool
- gait disturbance
- seizures and/or myoclonic jerks
- swallowing dysfunction
- sleep changes (eg day-night reversal, daytime fatigue)
- altered appetite and thirst
- personality or psychological changes

- depressed mood
- aggressiveness
- paranoia
- compulsiveness
- loss of interest in activities

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

Does the 23 year old woman mentioned at the beginning of this post have AD? It would certainly not be the first thing I would consider. The youngest person we diagnosed with AD was about 35 years old when his symptoms began. While there are individuals that will fall outside the usual age range for developing AD, 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 who don't have AD.

Also, not to sound like a broken record, but for further information, please see both of our books:

The Guide to Good Health
