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How Down syndrome may help unravel Alzheimer's puzzle

BY LINDA CARROLL

Scientists have known for decades that people with Down syndrome were at increased risk of developing Alzheimer's disease, but they didn't know why. Some researchers now believe that understanding the connection between the two conditions might help us unravel the Alzheimer's puzzle and point towards therapies that might slow, or even halt, the dreaded disease.

"It's a tantalizing and provocative question: Do people with Down syndrome hold the key to the mystery of Alzheimer's development?" Dr. Brian Skotko, co-director of the Down Syndrome Program at the Massachusetts General Hospital in Boston, said in a telephone interview. "And what can we learn from those with Down syndrome that will benefit the rest of the population?"

Not only do more people with Down syndrome develop Alzheimer's, but they also develop it at a much younger age. By age 40, a full 40 percent of people with Down syndrome will develop the disease, and by age 50 that rises to 50 percent, Skotko told TODAY's Maria Shriver.

While not everyone with Down syndrome develops dementia, all develop changes in their brains that are found in Alzheimer's patients – plaques made of a sticky protein called amyloid-beta that gunk up the spaces between nerve cells in the brain. Those plaques start to develop in people with Down syndrome at a very early age.

As it turns out, the precursor protein for amyloid-beta is encoded on the 21st chromosome, which happens to be the very chromosome that people with Down syndrome get an extra copy of, says Dr. Cindy Lemere, an associate professor of neurology at the Harvard Medical School and Brigham and Women's Hospital.

One of the interesting questions to be answered is why 50 percent of those with Down syndrome don't ever develop dementia even though they do develop plaques, Skotko said.

Scientists don't need to know the answer to that question to start looking for therapies to try to treat Alzheimer's.

"We've learned that prevention and treatment in the earliest stages is probably our best way to battle this disease," Lemere told TODAY. "And we know that everybody with Down syndrome will eventually develop Alzheimer's disease – or at least the changes in the brain. So we know that this is now another population where we can perhaps go in and test therapies very early in the disease as a prevention."

Skotko is currently finishing up the last of the paperwork for a clinical trial of an experimental therapy in Down syndrome volunteers. The trial will look at the impact of scyllo-inositol, a drug developed by the Elan Corporation that is designed to block plaque formation and to pump up the levels of a compound that appears to nourish cells of all sorts.

As a possible side effect, the drug might also improve the intellectual functioning of those with Down syndrome, Skotko says. The hypothesis is that the amyloid-beta plaques might be the cause of poor memory and learning difficulties experienced by people with Down syndrome.

Word has already gotten out about the upcoming trial, Skotko said. "Our phones have been ringing off the hook with parents who are really interested in learning about the clinical trials," he told Shriver. "Some parents say, 'If there is anything I could give my son or daughter that might improve memory, improve their cognition, I'm all for it.'"

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But other parents, as well as people who have Down syndrome, aren't as enthusiastic about drugs that might not only improve intellectual abilities, but might also have unknown and unpredictable effects on personality, among other things.

That's something Skotko understands intimately. His sister, Kristin, has Down syndrome and is the one who inspired him to get into research. His sister and the rest of the family are currently on the fence about the trials.

"I think it's an individual decision," Skotko told Shriver. "It's going to be a matter of whether or not she wants to be part of the experiments, of the clinical trials, before it's proven to be effective."

That rings true to Andy Majewski, whose son Ben has Down syndrome – and a job meeting and greeting patients and families at the Down Syndrome Program at Massachusetts General. Right now, Ben, Andy, and the rest of the family are mulling over the possible impact of the drug.

"We're struggling with it because, quite frankly, we think Ben is perfect," Andy Majewski told Shriver. "So we don't look for any changes in him. But the prospect of Alzheimer's makes you think a bit more about, if there's a potential cure, and this can unlock the code to Alzheimer's, we have to think about it a little more carefully.

"Ben is his own self-advocate. Ben has a say in it. And so, in this case, we would have to spend more time together as a family really understanding what are the risks, what are the potential rewards. At the end of the day it's an individual family choice, no different than it would be for any other kind of clinical trial."

1. Explain the proposed genetic connection between Down Syndrome and Alzheimer's Disease.

2. Imagine you had a child with Down Syndrome. If the clinical trial is successful and the new drug could improve the mental function of your child, would you want them to have it? Why or why not?

3. Why do you think 50% of people with Down syndrome don't ever develop dementia even though they do develop amyloid-beta plaques?

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