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Alzheimer's Disease in People With Down Syndrome

Many but not all people with Down syndrome develop <u>Alzheimer's disease</u> when they get older.

People with Down syndrome are born with an extra copy of chromosome 21, which carries a gene that produces a specific protein called amyloid precursor protein (APP). Too much APP protein leads to a buildup of protein clumps called beta-amyloid plaques in the brain. The presence of beta-amyloid plaques is one of the hallmarks of Alzheimer's disease.

By age 40, most people with Down syndrome have these plaques, along with other protein deposits, called tau tangles, which cause problems with how brain cells function and increase the risk of developing Alzheimer's symptoms. Estimates



suggest that 50% or more of people with Down syndrome will develop dementia due to Alzheimer's disease as they age.

This type of Alzheimer's in people with Down syndrome is not passed down genetically from a parent to a child.

Down Syndrome and Alzheimer's Research

Scientists are working hard to understand why some people with Down syndrome develop <u>dementia</u> while others do not. They want to know how Alzheimer's disease begins and progresses so they can develop drugs or other treatments that can stop, delay, or even prevent the disease process. They are also hopeful that research on Alzheimer's and Down syndrome will not only help those with both conditions, but also may lead to effective treatments for all people with Alzheimer's. Research in this area includes:

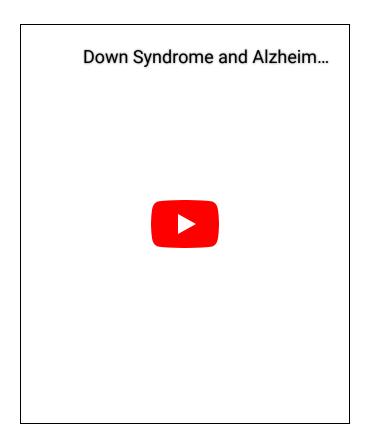
- Basic studies to improve understanding of the genetic and biological causes of brain abnormalities that lead to Alzheimer's
- Observational research to measure cognitive changes in people over time
- Studies of biomarkers (biological signs of disease), brain scans, and other tests that may help diagnose
 Alzheimer's even before symptoms appear and show brain changes as people with Down syndrome age
- Clinical trials to test treatments for dementia in adults with Down syndrome

The <u>Alzheimer's Biomarkers Consortium - Down Syndrome (ABC-DS)</u> is a National Institutes of Health (NIH)-funded research initiative to find biomarkers that indicate Alzheimer's disease is developing or progressing, and to track the Alzheimer's process in people with Down syndrome.

Alzheimer's and Down Syndrome Research Highlight

One ABC-DS <u>study</u> found disruptions in the way cells process and break down energy in people with Down syndrome who had Alzheimer's or mild cognitive impairment. Researchers measured these metabolic changes in the blood and found that they increased based on disease stage. Findings suggest that measuring such metabolic factors could be a potential way to track the disease.

In 2018, NIH launched the <u>INCLUDE (INvestigation of Co-occurring conditions across the Lifespan to Understand Down syndrome)</u> project to address health and quality-of-life needs for people with Down syndrome. Through INCLUDE-supported studies, researchers are exploring connections between Down syndrome and other conditions including Alzheimer's, autism, cataracts, celiac disease, congenital heart disease, and diabetes.



Alzheimer's Disease and Down Syndrome

Bob volunteers for Alzheimer's clinical trials because his daughter has Down syndrome. Volunteering in clinical trials can help researchers understand why people with Down syndrome often develop Alzheimer's disease.

Hear Bob's Story

Learn About Alzheimer's Disease Research Opportunities

- Join <u>DS-Connect</u>, a voluntary, confidential, online registry from the NIH *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD). You, your legally authorized representative, or your guardian can submit information about your health and choose to be contacted about research opportunities, if desired. Email DS-Connect for more information and to sign up.
- Learn more about the NIH INCLUDE (INvestigation of Co-occurring conditions across the Lifespan to Understand Down syndromE) project.

- Learn about the <u>Alzheimer's Clinical Trials Consortium Down Syndrome (ACTC-DS)</u> and the <u>Trial Ready-Cohort for Down Syndrome (TRC-DS)</u>, which is a study enrolling people who are willing and ready to participate in current and future clinical trials.
- Learn more about <u>participating in Alzheimer's disease research</u> and search the <u>Alzheimers.gov Clinical Trials</u> Finder.
- · Visit NIH's "Clinical Research Trials and You."

You may also be interested in

- Exploring how Alzheimer's disease is diagnosed
- Learning ways to <u>deal with agitation and aggression in Alzheimer's disease</u>
- Watching a video about genetics and Alzheimer's disease

For More Information About Down Syndrome and Alzheimer's

Down Syndrome and Alzheimer's Disease Resources

- Alzheimer's Disease in People With Down Syndrome Fact Sheet (NIA)
- <u>Aging and Down Syndrome: A Health & Well-Being Guidebook</u> (PDF, 8.0M) (National Down Syndrome Society).
 Also available in <u>Spanish</u> (PDF, 8.0M)
- Alzheimer's Disease and Down Syndrome: A Practical Guide for Caregivers (PDF, 3.3M) (National Down Syndrome Society). Also available in <u>Spanish</u> (PDF, 3.1M)
- Alzheimer's Disease and Down Syndrome: Overview (National Down Syndrome Society)
- <u>Down Syndrome and Alzheimer's Disease</u> (Alzheimer's Association)

Down Syndrome Research and Resources

- Down Syndrome Biobank Consortium
- <u>Down Syndrome: Overview</u> (NICHD)
- DS-Connect The Down Syndrome Registry (NIH)
- INCLUDE Project: Investigation of Co-occurring conditions across the Lifespan to Understand Down syndrome
 (NIH)
- Global Down Syndrome Foundation
- LuMind IDSC Foundation
- Down Syndrome Medical Interest Group

NIA Alzheimer's and related Dementias Education and Referral (ADEAR) Center

800-438-4380

adear@nia.nih.gov

www.nia.nih.gov/alzheimers

The NIA ADEAR Center offers information and free print publications about Alzheimer's and related dementias for families, caregivers, and health professionals. ADEAR Center staff answer telephone, email, and written requests and make referrals to local and national resources.

Alzheimers.gov

www.alzheimers.gov

Explore the Alzheimers.gov website for information and resources on Alzheimer's and related dementias from across the federal government.

This content is provided by the NIH National Institute on Aging (NIA). NIA scientists and other experts review this content to ensure it is accurate and up to date.

Content reviewed: November 30, 2020

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