

# Blood test detects Alzheimer's in people with Down syndrome

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Around 80% of people with Down syndrome develop Alzheimer's disease, often when they are between 40 and 50 years old. A study led by Lund University in Sweden has shown that a simple blood test can detect

Alzheimer's disease in people with Down syndrome with a high degree of certainty. The findings are important for several reasons, not least the ability to make a correct diagnosis without invasive procedures. The study was recently published in *JAMA Neurology*.

This is, so far, the only relatively large study in the world on Down syndrome in which a blood biomarker is compared with PET imaging results to see if people have the significant accumulations of the proteins in the brain that define Alzheimer's disease. The study involved 300 people with Down syndrome, 40% of whom showed signs of the onset of Alzheimer's disease.

"With a [simple blood test](#), we were able to detect both tau and amyloid pathologies, the disease-related changes in the brain that indicate whether a person has Alzheimer's or not, with over 90% certainty. We were even able to diagnose those who had not yet developed clear [cognitive impairment](#) due to Alzheimer's disease," says Oskar Hansson, professor of Neurology at Lund University and senior consultant at Skåne University Hospital.

In 2020, Oskar Hansson and his research colleagues announced their major breakthrough in Alzheimer's diagnostics: that a biomarker in the blood, phosphorylated tau (P-tau217) can detect Alzheimer's disease as early as 20 years before memory problems become apparent and, most importantly, distinguish Alzheimer's disease from other forms of dementia with about 95% accuracy. Clinical studies are now underway at 25 health centers in Sweden, that include both cognitive assessments and measurements of P-tau217 in blood.

"We used the same [blood biomarker](#) in this study. Many people are unaware that Alzheimer's disease occurs much more frequently and at an earlier age in people with Down syndrome. It is also more complicated to diagnose Alzheimer's in a person with Down syndrome, as there is a

pre-existing intellectual disability that makes it more difficult to detect cognitive impairment, and it requires the patient to agree to invasive procedures such as spinal fluid tests. It is therefore of particular importance that we find a simple diagnostic method," says Oskar Hansson.

The reason people with Down syndrome develop Alzheimer's disease at a comparatively high rate is that they have an extra chromosome, three copies of chromosome 21 instead of two. The gene for the [amyloid precursor protein](#) (APP) that is cleaved to produce amyloid, is located on chromosome 21.

"Down syndrome produces more of the APP protein and thus results in a significantly increased risk of amyloid aggregates, which in turn lead to tau aggregates," says Shorena Janelidze, researcher at Lund University.

The study obtained both [blood samples](#) and PET scan results for people with Down syndrome, providing a unique opportunity to investigate whether the biomarker could also serve as a diagnostic marker for people with Down syndrome.

"Our results show that P-tau217 works just as well as a blood marker of Alzheimer's disease for persons with Down syndrome as it does for others and that other blood markers are not needed, Ptau217 is sufficient. The next step is to evaluate the performance of this biomarker in [clinical practice](#) and use it to improve [clinical trials](#) evaluating drugs targeting Alzheimer's disease in people with Down syndrome," concludes Janelidze.

**More information:** Shorena Janelidze et al, Detection of Brain Tau Pathology in Down Syndrome Using Plasma Biomarkers, *JAMA Neurology* (2022). [DOI: 10.1001/jamaneurol.2022.1740](https://doi.org/10.1001/jamaneurol.2022.1740)

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