

# Study finds Alzheimer's disease link in eyes of children with Down syndrome

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A team of researchers has discovered that the protein that forms plaques in the brain in Alzheimer's disease also accumulates in the eyes of people with Down syndrome. The new findings in Down syndrome show that the toxic protein, known as amyloid- $\beta$ , that causes Alzheimer's pathology in the brain also leads to distinctive cataracts in the eyes. The discovery is leading the researchers to develop an innovative eye test for early detection of Alzheimer's pathology in both disorders.

The research, led by Lee E. Goldstein, M.D., Ph.D., associate professor at Boston University School of Medicine and the Boston University [Alzheimer's Disease](#) Center, and Juliet A. Moncaster, Ph.D., associate director of the Molecular Aging & Development Laboratory, also at Boston University, was presented at the annual meeting of the Association for Research in Vision and Ophthalmology in Fort Lauderdale, Florida and reported in the May 20 issue of *PLoS One*. The research included investigators at the Brigham & Women's Hospital; Massachusetts Eye and Ear Infirmary; Massachusetts General Hospital; Harvard Medical School; Rush University Medical Center; Children's Hospital Boston, and the University of Washington, Seattle.

"People with [Down syndrome](#) develop symptoms of Alzheimer's-type dementia often by the age of 30," said Goldstein, senior corresponding author on the *PLoS One* article. "This is because they have an extra copy of a key Alzheimer's gene that leads to increased amyloid- $\beta$  accumulation in the brain. We discovered that this same protein starts to accumulate very early in the lens of the eye, even in children, " explained

Goldstein.

"The lens provides a window to the brain," said Moncaster, co-lead author of the study. "The lens can't clear protein deposits the way the brain does. Our findings show that the same amyloid- $\beta$  protein that aggregates in the brain also accumulates in the lens and leads to these unusual cataracts in Down syndrome."

"The results are striking," added David G. Hunter, M.D., Ph.D., Ophthalmologist-in-Chief at Children's Hospital Boston and Vice Chairman of the Department of Ophthalmology at Harvard Medical School. "We have known that these cataracts are prevalent in people with Down syndrome and are sometimes seen at birth, but we never knew how they were related to the disorder—now we know," said Hunter. "These distinctive cataracts appear only in people with advanced Alzheimer's disease and much earlier in Down syndrome."

According to the National Down Syndrome Society and the National Institute of Child Health & Development, Down syndrome is the most common chromosomal disorder and the leading genetic cause of intellectual disability. In the US, one of every 800 newborns and more than 400,000 people have Down syndrome. Although the disease is incurable, life expectancy for people living with Down syndrome has increased dramatically in recent decades — from 25 in 1983 to 60 today.

"We are developing an eye scanner to measure amyloid- $\beta$  in the lens," said Goldstein. "This approach may provide a way for early detection and monitoring of related pathology in the brain. Effective treatments for the [brain](#) disease in Down syndrome and Alzheimer's disease are on the horizon, and early detection is the key for successful intervention," he said. "The path to effective treatment is what drives our research."

Provided by Boston University Medical Center

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