Scientists Identify a New Form of Early-Onset Alzheimer's

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Evidence shows that specific gene mutations can sometimes trigger early-onset Alzheimer's disease (AD), the signs and symptoms of which usually appear between ages 30 and 60. Now findings published in the journal Science Translational Medicine show that when amyloid precursor protein (APP)—one of a trio of genes responsible for this type of dementia—lacks a chain of certain amino acids this can trigger a rare, accelerated and severe type of Alzheimer's, reports MedicalXpress.com.

For the study, a multinational team of researchers collaborated with scientists from Uppsala University in Sweden. Scientists used genetic analysis, structural biological research, amino acid and protein chemistry studies and mass spectrometry—a technique used to identify chemical substances—to evaluate amyloid protein found in the brain tissue of the members in one family.

Initially, two siblings, age 40 and 43, respectively, and a cousin, also in his 40s, visited the clinic at Uppsala University Hospital after they experienced difficulty speaking and an inability to perform simple mathematical calculations. The relatives also suffered from memory problems, a lost sense of direction and mental acuity. After examining their brain scans, doctors diagnosed the three with early-onset AD.

Interestingly, 20 years earlier, the siblings' father was also in his 40s when he experienced similar symptoms and went to the same clinic for treatment.

"Affected individuals have an age at symptom onset in their early forties, and suffer from a rapidly progressing disease course," explained María Pagnon de la Vega, PhD, the lead author of the study.

The modern technology used to check for genetic mutations responsible for this aggressive form of early-onset AD showed that each family member lacked a series of six amino acids found in APP genes. Scientists called the insufficiency, which causes sticky clumps of harmful amyloid beta plaque to form in the brain, "the Uppsala APP deletion."

This multiple amino acid deletion is the first such mutation that researchers have identified as a cause of early-onset Alzheimer's disease.

Interestingly, previous research published in the journal Neurology, which focused on a large

African-American family, found a different kind of APP mutation associated with the early onset of Alzheimer's. In this group of relatives, however, the condition struck individuals in their early 30s and was accompanied by episodic seizures and myoclonus, an involuntary twitching of the muscles.

For more news about AD, read "FDA Approves Controversial Alzheimer's Treatment" and "New Blood Test May Predict Alzheimer's Disease."
