

Common nutritional supplement offers promise in treatment of unique form of autism with epilepsy

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An international team of researchers, led by scientists at the University of California, San Diego and Yale University schools of medicine, have identified a form of autism with epilepsy that may potentially be treatable with a common nutritional supplement.

The findings are published in the September 6, 2012 online issue of *Science*.

Roughly one-quarter of [patients](#) with autism also suffer from epilepsy, a brain disorder characterized by repeated seizures or convulsions over time. The causes of the epilepsy are multiple and largely unknown. Using a technique called exome sequencing, the UC San Diego and Yale scientists found that a [gene mutation](#) present in some patients with autism speeds up metabolism of certain amino acids. These patients also suffer from epileptic seizures. The discovery may help physicians diagnose this particular form of autism earlier and treat sooner.

The researchers focused on a specific type of amino acid known as branched chain amino acids or BCAAs. BCAAs are not produced naturally in the human body and must be acquired through diet. During periods of starvation, humans have evolved a means to turn off the metabolism of these amino acids. It is this ability to shut down that [metabolic activity](#) that researchers have found to be defective in some autism patients.

"It was very surprising to find mutations in a potentially treatable [metabolic pathway](#) specific for autism," said senior author Joseph G. Gleeson, MD, professor in the UCSD Department of Neurosciences and Howard Hughes Medical Institute investigator. "What was most exciting was that the potential treatment is obvious and simple: Just give affected patients the naturally occurring

[amino acids](#) their bodies lack."

Gleeson and colleagues used the emerging technology of exome sequencing to study two closely related families that have children with [autism spectrum disorder](#). These children also had a history of seizures or abnormal electrical [brain wave activity](#), as well as a mutation in the gene that regulates BCAAs. In exome sequencing, researchers analyze all of the elements in the genome involved in making proteins.

In addition, the scientists examined cultured neural stem cells from these patients and found they behaved normally in the presence of BCAAs, suggesting the condition might be treatable with nutritional supplementation. They also studied a line of mice engineered with a mutation in the same gene, which showed the condition was both inducible by lowering the dietary intake of the BCAAs and reversible by raising the dietary intake. Mice treated with BCAA supplementation displayed improved neurobehavioral symptoms, reinforcing the idea that the approach could work in humans as well.

"Studying the animals was key to our discovery," said first author Gaia Novarino, PhD, a staff scientist in Gleeson's lab. "We found that the mice displayed a condition very similar to our patients, and also had spontaneous [epileptic seizures](#), just like our patients. Once we found that we could treat the condition in mice, the pressing question was whether we could effectively treat our patients."

Using a [nutritional supplement](#) purchased at a health food store at a specific dose, the scientists reported that they could correct BCAA levels in the study patients with no ill effect. The next step, said Gleeson, is to determine if the supplement helps reduce the symptoms of epilepsy and/or autism in

humans.

"We think this work will establish a basis for future screening of all patients with autism and/or epilepsy for this or related genetic mutations, which could be an early predictor of the disease," he said. "What we don't know is how many patients with [autism](#) and/or epilepsy have mutations in this gene and could benefit from treatment, but we think it is an extremely rare condition."

Provided by University of California - San Diego

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