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Autism discovery paves way for early blood test and therapeutic options

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Researchers at the JC Self Research Institute of the Greenwood Genetic Center (GGC), along with collaborators from Biolog, Inc. in California, have reported an important discovery in the understanding of autism which was published this week in *Molecular Autism*.

The study, led by GGC's Director of Research, Charles Schwartz, PhD and Staff Scientist, Luigi Boccuto, MD, found that individuals with autism spectrum disorders (ASDs) showed significantly decreased metabolism of the amino acid L-tryptophan when compared to both typical controls and individuals with other neurodevelopmental disorders . Cells from individuals with autism metabolized L-tryptophan at a decreased rate whereas cells from individuals without autism did not show this change.

Researchers also measured the expression of genes that are known to be involved in L-tryptophan metabolism in a small subset of patients with autism and found they also expressed some of the genes at lower levels than those without autism.

"The important and immediate implication of this work is the development of a simple, early blood screening test for autism by measuring the metabolism of L-tryptophan using Biolog's technology," shared Dr. Boccuto. Biolog's assay method, called Phenotype MicroArray technology, allows researchers to measure the ability of cells to generate energy from various biochemical nutrients, including L-tryptophan.

Currently there are no laboratory tests that can accurately diagnose ASDs, which are estimated to affect 1 in 50 school-aged children in the US. Current diagnosis depends upon a developmental evaluation and parent interviews and can often not be made prior to 2-3 years of age. "A screening, and eventually, a diagnostic blood test for autism would be of immense value to families," explained Dr. Schwartz. "An early, accurate diagnosis is key to providing effective and timely therapies for these patients and their families."

Dr. Boccuto added, "We also see tremendous potential that these findings will aid in our understanding of the molecular and metabolic bases of autism. Once we have a clear vision of what has gone awry within the tryptophan metabolism pathways, we can develop therapies to target and correct those problems at the biochemical level."

L-tryptophan is one of twenty amino acids used by cells to make protein. It is one of eight amino acids that cannot be made by the body, so it must be obtained from the diet. More importantly, L-tryptophan plays an important role in brain development and function as it is the precursor of key neurochemicals such as serotonin and melatonin which have already been linked to behavioral and neurodevelopmental problems.

"This discovery leads us toward a possible unifying biochemical mechanism for ASDs which could ultimately lead to a treatment," shared Dr. Schwartz. "Now that we have additional evidence that the features of ASDs may be related to the metabolic pathways involving L-tryptophan, we can focus further studies on determining at what point along those pathways the disruption occurs, which may vary from one patient to another. With treatments that target various points along the pathway, a modality that works for one patient may not work for another."

GGC's autism research has been supported by funds from the South Carolina Department of Disabilities and Special Needs. Additional

funding has been obtained from the National Institutes of Health to explore transitioning the research finding into a simple blood test for autism. Drs. Schwartz and Boccuto are currently evaluating the tryptophan metabolism in fresh blood samples from patients with ASDs and controls, utilizing customized Biolog plates.

"We are thrilled that Biolog's technology helped Dr. Schwartz in his pioneering research and that it has led to this breakthrough discovery," said Barry Bochner, PhD, CEO at Biolog, Inc.

Provided by Greenwood Genetic Center