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Title: Study Discovers Statistically Significant Link Between Abnormally Low Cholesterol Levels and Autism Spectrum Disorders

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Finding Leads Kennedy Krieger Researchers Down New Road in Autism Research
 BALTIMORE, M.D. -- September 7, 2006 -- Autism spectrum disorders (ASD) will be diagnosed in more children this year than AIDS, diabetes and cancer combined, yet researchers and physicians can identify medical causes in only 10 percent of cases. But, a study published in the [*American Journal of Medical Genetics Part B (Neuropsychiatric Genetics)*] found that a small subgroup of children with ASD have abnormally low cholesterol levels (hypocholesterolemia), leading researchers to believe cholesterol may play a role in the cause of some cases of the disorder. The children's low cholesterol levels were apparently due to a limited ability to make cholesterol.

Nineteen of the 100 children who participated in the study were found to have total cholesterol levels below 100 mg/dL, which is lower than that found in 99 percent of children. The average cholesterol level for children between 4 and 19 years of age is 165 mg/dl, as determined by the Center for Disease Control's National Health and Nutrition Examination Survey (2006).

The study authors found evidence that the low cholesterol levels were caused by a reduced ability of the body to naturally produce cholesterol, and not by inadequate amounts of cholesterol in the diet or gastrointestinal problems that interfere with cholesterol absorption, two of the more common causes of low blood cholesterol levels. Of the 19 individuals in the low cholesterol group, 13 (68%) met criteria for an autism diagnosis and 6 (32%) met criteria for a different disorder on the autism spectrum, such as Asperger syndrome or pervasive developmental disorder not otherwise specified (PDD-NOS).

"We know that people can tolerate having low cholesterol, so we suspect that deficiencies in cholesterol combined with mutations to a specific gene may have resulted in autism spectrum disorders in these children," said lead author Elaine Tierney, MD, Director of the Autism Metabolic Research Program at the Kennedy Krieger Institute in Baltimore. "Our next steps are to determine if other abnormalities of cholesterol metabolism can be risk factors for the development of autism."

The study examined blood samples from 100 children with ASD who met four criteria: 1) member of a family with two or more individuals with ASD; 2) over two years of age; 3) abnormally slow attainment of a subset of developmental milestones; and 4) assessment utilizing a standardized autism

interview. Blood samples were drawn from the Autism Genetic Resource Exchange (AGRE), a blood collection repository. Although only a very small proportion of children with autism come from families with two or more affected children, these children are much more likely to have a genetic cause of autism and less likely to have autism due to an environmental cause or early brain injury.

"This study underscores the critical importance of families participating in blood collection repositories," said Dr. Gary Goldstein, President and CEO of the Kennedy Krieger Institute. "I believe these repositories hold great promise in helping researchers explore causes and potential treatments for autism spectrum disorders, such as a possible nutritional intervention for some of these children with cholesterol deficiencies."

Researchers at the Kennedy Krieger Institute, in collaboration with the National Institutes of Health and AGRE, the organization that provided the 100 blood samples tested in this study, have now begun analyzing the genes of the children with low cholesterol. These children's genes are being compared to their parents' to determine if there are common mutations affecting cholesterol metabolism being passed from parent to child. These same families have also performed behavioral and IQ testing with AGRE, allowing data to be grouped and analyzed based upon the outcomes of the gene research. This study was supported by funding from Cure Autism Now and the Smith-Lemli-Opitz/RSH Foundation.

About Autism Spectrum Disorders (ASD)

Autism is the fastest growing developmental disorder in the United States. Today, 1 in 166 individuals is diagnosed with the disorder, which occurs in all racial, ethnic, and social groups and is four times more likely to strike boys than girls. Deficits in social interaction, limited verbal and nonverbal communication and repetitive behaviors and obsessions characterize children with ASD, whose formal diagnosis may be autism, or a milder disorder on the spectrum such as Asperger syndrome or pervasive developmental disorder not otherwise specified (PDD-NOS). For most affected individuals, autism is a serious and lifelong disorder. While there have been notable advances in ASD research over the last 10 years, the cause of autism in most children remains unknown. Continued research is crucial to understanding all causes of ASD and allowing the earliest detection and intervention for affected families. Previous studies have shown that early identification and treatment can lead to improved outcomes in individuals with ASD.

SOURCE: Kennedy Krieger Institute

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