Medical Xpress

Mental deficiency: Researchers identify gene mutations that affect learning, memory in children

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Mental deficiency is the most frequently occurring, yet least understood handicap in children. Even a mild form can lead to social isolation, bullying and require assistance with simple tasks. The most common variety, non-syndromic mental deficiency (NSMD), is defined as affecting an otherwise normal looking child. With few physical clues in affected children to point researchers towards candidates to study, progress in identifying genetic causes of NSMD has been very slow. Yet that is beginning to change.

Jacques L. Michaud, a geneticist at the Sainte-Justine University Hospital Research Center and the Centre of Excellence in Neuromics of the Université de Montréal, has led a multidisciplinary team which has identified mutations in a novel gene in children with NSMD. Their study is published in today's issue of the *New England Journal of Medicine* and includes collaborators from McGill University in Canada, the National Institute of Mental Health and the Nathan S Kline Institute in the U.S. and the Université Paris Descartes in France.

"NSMD is a disorder that has many causes," says Dr. Michaud. "By linking this gene to one kind of NSMD, we better understand the causes and we can work towards a way of identifying and treating this incapacitating condition".

The identified mutations affect the function of SYNGAP1, a gene that

codes for a protein involved in the development and function of the connections between brain cells, also called synapses. The disruption of this gene has been shown to impair memory and learning in mice.

A new approach

Dr. Michaud's research team hypothesized that new mutations that arise in children - while not present in their parents - may represent a common cause of mental deficiency. "Several observations indicate that new mutations are a frequent cause of neurodevelopmental disorders, but their identification has been difficult because it requires the study of a large fraction of genes, which represents a challenging task," says Dr. Fadi F. Hamdan, first author of the study.

In order to identify these new mutations, the team took advantage of the platform developed by the Synapse to Diseases consortium, based in Montreal, to study 500 synaptic genes in a group of children with unexplained mental deficiency. The team found that three percent of their subjects had new deleterious mutations in the SYNGAP1 gene.

"This discovery illustrates the power of novel technologies that allow researchers to study hundreds of genes in large groups of individuals, and provides validation for the use of such an approach for the exploration of neurodevelopmental disorders," says Dr. Guy A. Rouleau, Director of Sainte-Justine Research Center and Head of the Synapse to Diseases consortium.

Impact of the discovery

Children with mutations in SYNGAP1 show strikingly similar forms of NSMD, with delays in their language and mental development and, in some cases, a mild form of epilepsy. Now that these SYNGAP1

mutations have been linked to NSMD, diagnostic tests can be offered to children with NSMD, and adapted strategies of learning can be developed. Moreover, because of the wealth of knowledge about the function of SYNGAP1, it may also be possible to design targeted pharmacological therapies that would aim at improving cognition and associated complications such as epilepsy.

Source: University of Montreal

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