Press Release

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Genomic Region Associated with Autism Plays Role in Specific Cognitive Functions

Reports new study in Biological Psychiatry

Philadelphia, PA, July 18, 2016 – A new study in *Biological Psychiatry* reports that variations in 16p11.2, a region of the genome associated with risk of autism spectrum disorder (ASD), have distinct effects on cognition. The findings highlight the diversity of people with ASD.

Extra or missing copies of genetic material in a small region of the genome in chromosome 16, designated 16p11.2, increases the risk of autism spectrum disorders. Known as duplications or deletions, these alterations in the 16p11.2 genomic region are also associated with intellectual disability.

In the first study to look at the effect of both duplications and deletions in 16p11.2 on specific cognitive domains, senior author Dr. Sébastien Jacquemont, from the University of Montreal in Canada, and a large research team assessed the effects of these variations in 62 deletion carriers, 44 duplication carriers, and 71 controls from within the same families. According to Jacquemont, determining the effect of these alterations can only be performed through family studies. The researchers used neuropsychological tests to assess overall cognitive functioning, fine motor skills, language, memory, and executive functions.

After accounting for the lower IQ associated with 16p11.2 variations, differences in specific cognitive domains emerged. Deletion carriers had difficulty with phonology, reading fluency, fine motor skills, and verbal and motor inhibition. Duplication carriers outperformed controls with the same IQ on tasks of verbal memory, executive functions, and phonological skills. The authors suggest this is reminiscent of the complex and conflicting association between language impairment and autism.

“These data suggest that copy number variants may generally increase risk for intellectual disability and autism, but that the particular nature of the genetic alteration may have specific functional consequences for brain and behavior,” said Dr. John Krystal, Editor of *Biological Psychiatry*.

“Genomic variants associated with ASD may be associated with very different cognitive alterations and profiles,” said Jacquemont, “and we may learn something on the developmental mechanisms involved in ASD by focusing on the cognitive comorbidities.” He added that generating similar data across other regions of the genome will be essential to understand the impact of different variants on development.

The findings of the study may help better inform the type of intervention that patients will benefit from most; the authors write that the use of visuospatial processes when learning may help patients who carry a deletion, whereas verbal methods may improve learning strategies in patients who carry a duplication.

Notes for editors

The article is "The Number of Genomic Copies at the 16p11.2 Locus Modulates Language, Verbal Memory, and Inhibition," by Loyse Hippolyte, Anne M. Maillard, Borja Rodriguez-Herreros, Aurélie Pain, Sandra Martin-Brevet, Carina Ferrari, Philippe Conus, Aurélien Macé, Nouchine Hadjikhani, Andres Metspalu, Anu Reigo, Anneli Kolk, Katrin Männik, Mandy Barker, Bertrand Isidor, Cédric Le Caignec, Cyril Mignot, Laurence Schneider, Laurent Mottron, Boris Keren, Albert David, Martine Doco-Fenzy, Marion Gérard, Raphael Bernier, Robin P. Goin-Kochel, Ellen Hanson, LeeAnne Green Snyder, 16p11.2 European Consortium, Simons Variation in Individuals Project Consortium, Franck Ramus, Jacques S. Beckmann, Bogdan Draganski, Alexandre Reymond, and Sébastien Jacquemont (doi: 10.1016/j.biopsych.2015.10.021). It appears in *Biological Psychiatry*, volume 80, issue 2 (2016), published by Elsevier.

Copies of this paper are available to credentialed journalists upon request; please contact Rhiannon Bugno at +1 214 648 0880 or biol.psych@utsouthwestern.edu. Journalists wishing to interview the authors may contact Sébastien Jacquemont at sebastien.jacquemont@umontreal.ca.

The authors' affiliations, and disclosures of financial and conflicts of interests are available in the article.

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The journal publishes novel results of original research which represent an important new lead or significant impact on the field, particularly those addressing genetic and environmental risk factors, neural circuitry and neurochemistry, and important new therapeutic approaches. Reviews and commentaries that focus on topics of current research and interest are also encouraged.

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