Biomarker progress offers hope for early autism spectrum disorder detection

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Autism spectrum disorders (ASD) are neurodevelopmental disorders typically characterized by difficulties in social interactions and delayed or abnormal language development. Although ASD reportedly affects 1 in 88 people in the United States, to date there have been no distinctive biomarkers to diagnose the disease. In a special themed issue of *Disease Markers*, investigators report on the current understanding of ASD genetics and the possibilities of translating genetic research toward biomarker development in ASD.

"Although some individuals with ASD are highly functional, many are severely impaired and require permanent care. The significant level of impairment combined with the fact that no specific therapy is yet available for ASD, make ASD a devastating illness for patients and families, and a heavy financial burden for the healthcare system," says guest editor, Irina Voineagu, MD, PhD, RIKEN Omics Science Center, Yokohama, Japan. "The most effective intervention for ASD has proven to be early behavioral therapy. Thus the identification of biological markers for ASD, allowing very early detection, even before the onset of symptoms, would be of tremendous value."

Five articles comprise this comprehensive issue, providing an overview of ASD genetic models, an exploration of several key emerging concepts in understanding ASD's molecular basis, and discussion of current biomarker development, focusing on genomic data.

Following an introduction by Voineagu, Yuri Bozzi and colleagues

review the phenotype characteristics of currently available mouse models of ASD. Carmen Panaitof then discusses the role of the songbird as an experimental model system for investigating the genetic basis of human language and its ASD-related impairments. Michael Bowers and Genevieve Konopka further explore language deficits and provide new evidence for the role of the FOXP gene to regulate language. Alka Saxena, Dave Tang, and Piero Carninci focus on the functional roles of the gene MECP2, which is mutated in most cases of Rett syndrome, one of the ASDs.

A review rounding out the issue is "Subphenotype-Dependent Disease Markers for Diagnosis and Personalized Treatment of Autism Spectrum Disorders," by Valerie W. Hu, PhD, The George Washington University, School of Medicine and Health Sciences, Washington, DC, PhD, which discusses current progress toward identifying ASD biomarkers based on genome-wide data.

"Without genetic or molecular markers for screening, individuals with ASD are typically not diagnosed before the age of 2, with milder cases diagnosed much later," writes Dr. Hu. "Because early diagnosis is tantamount to early behavioral intervention, which has been shown to improve individual outcomes, an objective biomarker test that can diagnose at-risk children perinatally is a medical imperative."

Hu demonstrates the possibility and importance of developing ASD subtypes to help identify relevant disease markers, which can ultimately aid in developing specific targeted therapies.

Voineagu concludes, "It is exciting times for genetic research and although the phenotypic and genetic heterogeneity of ASD often seem to be a daunting conundrum, well-defined diagnostic criteria, larger cohort sizes for genetic studies and integrative approaches of genomic and epigenomic data already delineate a promising avenue for elucidating the

mechanisms of ASD."

More information: "Autism: From Genetics to Biomarkers." Irina Voineagu. *Disease Markers*, Volume 33, Issue 5 (November 2012).

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