Multiparametric Diagnostic Biomarkers of Autism Spectrum Disorder a Contemporary Approach and Future Direction

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Abstract

Autism spectrum disorder (ASD) encompasses a heterologous group of neurodevelopmental disorders stemming from diverse combinations of genetic, metabolic, and environmental factors. Despite the many putative biomarkers known today, none of these has shown demonstrable effectiveness in screening and diagnosing patients before the onset of clinical symptoms or been introduced into clinical practice. Accurate identification of the pathophysiological pathways that underly ASD is important for diagnostic purposes as well as for understanding the specific molecular etiologies of the disease. Future biomarkers should be designed with the aims of (1) classifying ASD into categories based on treatable pathophysiologic pathways and (2) monitoring disease progression and treatment response. Given the complexity and heterogeneity of the disease process, achieving such aim will likely require designing therapeutically responsive,
multiparametric biomarker profiles that define each category. Multiparametric profiles have shown improved predictive power over individual biomarkers in the context of various disease conditions, including ASD. We will provide examples of multiparametric biomarker profiles and how they compare to individual biomarkers, summarize our group's work during the past decade in the field of ASD biomarkers, and discuss examples of the statistical modelling methodologies commonly used in the field.