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Biomarkers and Pathways in Autism Spectrum Disorder: An Individual Meta-Analysis Based on Proteomic and Metabolomic Data

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ABSTRACT

The utilization of biomarkers for the diagnosis and management of autism spectrum disorders (ASD) remains a relatively unexplored frontier in clinical practice. Proteomics and metabolomics are important tools for revealing key biomarkers and evaluating biological pathways in ASD. We conducted an individual meta-analysis to compare the consistency of biomarkers of ASD from central nervous system (brain and cerebrospinal fluid), circulatory system (blood), and non-invasive samples (urine, saliva, and faeces) and performed pathway enrichment analyses to identify pathways enriched in ASD. After screening 926 proteomics and 619 metabolomics articles, we collected data from 10 studies involving 940 differential proteins and 12 studies assessing a total of 748 differential metabolites. In brain tissue, blood, and urine of ASD cases and controls, flotillin-2 (FLOT2), apolipoprotein E (APOE), and EH domain-containing protein 3 (EHD3) exhibit differential expression, while vinculin (VCL) displays variations in saliva, blood, and urine. Similarly, in case-control studies, gelsolin (GSN) shows differential expression in brain tissue, saliva, and urine, and malate dehydrogenase 2 (MDH2) in brain tissue, blood, and saliva. Hippuric acid and salicyluric acid were simultaneously found in the brain, blood, urine, and faeces. In terms of pathways, glycolysis/gluconeogenesis, carbon metabolism, and glutathione metabolism were enriched in the brain as well as in saliva or urine. In our study, we identified six shared protein and two metabolic markers in central nervous system, circulatory system, and non-invasive samples, underscoring their potential value for ASD diagnosis and management, warranting further research.

Keywords: public health, psychology, screening biomarkers, child research