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ORIGINAL PAPER

A Prospective Study of Transsulfuration Biomarkers in Autistic Disorders

David A. Geier · Janet K. Kern · Carolyn R. Garver · James B. Adams · Tapan Audhya · Mark R. Geier

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Abstract The goal of this study was to evaluate transsulfuration metabolites in participants diagnosed with autism spectrum disorders (ASDs). Transsulfuration metabolites, including: plasma reduced glutathione (GSH), plasma oxidized glutathione (GSSG), plasma cysteine, plasma taurine, plasma sulfate, and plasma free sulfate among participants diagnosed with ASDs (n = 38) in comparison to age-matched neurotypical controls were prospectively evaluated. Testing was conducted using Vitamin Diagnostics, Inc. (CLIA-approved). Participants diagnosed with ASDs had significantly ($P < 0.001$) decreased plasma reduced GSH, plasma cysteine, plasma taurine, plasma sulfate, and plasma free sulfate relative to controls. By contrast, participants diagnosed with ASDs had significantly ($P < 0.001$) increased plasma GSSG relative to controls. The present observations are compatible with increased oxidative stress and a decreased detoxification capacity, particularly of mercury, in patients diagnosed with ASDs. Patients diagnosed with ASDs should be routinely tested to evaluate transsulfuration metabolites, and potential treatment protocols should be evaluated to potentially correct the transsulfuration abnormalities observed.

Keywords Heavy metal · Metabolic endophenotype · Sulfation · Sulfur

D. A. Geier
Institute of Chronic Illnesses, Inc., Silver Spring, MD, USA

D. A. Geier
CoMeD, Inc., Silver Spring, MD, USA

J. K. Kern · C. R. Garver
Autism Treatment Center, Dallas, TX, USA

J. K. Kern
University of Texas Southwestern Medical Center, Dallas, TX, USA

J. B. Adams
Arizona State University, Tempe, AZ, USA

T. Audhya
Vitamin Diagnostics, Cliffwood Beach, NJ, USA

M. R. Geier (✉)
The Genetic Centers of America, 14 Redgate Ct., Silver Spring 20905, MD, USA
e-mail: mgeier@comcast.net

Introduction

Autism spectrum disorders (ASDs) are prevalent neurodevelopmental disorders that affect an estimated 1 in 150 children in the US [1]. It has been observed that ASDs are characterized by impairments in social relatedness and communication, repetitive behaviors, abnormal movement patterns, and sensory dysfunction [2]. Further, common co-morbidity conditions often associated with ASDs include gastrointestinal disease and dysbiosis [3], autoimmune disease [4], and mental retardation [5].

In attempting to understand the underlying pathogenesis of ASDs, a considerable body of research has been conducted to evaluate potential candidate causal genes. Genetic studies, to date, have not uncovered genes of strong effect. It has recently been postulated that increasing rates of ASDs and less than 100% monozygotic concordance of ASDs support a more inclusive reframing of ASDs as a multisystem disorder with genetic influence and environmental contributors [6].

Research into the metabolic basis for ASDs has been relatively underutilized compared to other approaches.