



A Prospective Assessment of Porphyrins in Autistic Disorders: A Potential Marker for Heavy Metal Exposure

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(Submitted 24 June 2006; Revised 31 August 2006; In final form 31 August 2006)

Autism was recently associated with a urinary porphyrin pattern indicative of mercury toxicity in a large cohort of French children. The IRB of the Institute for Chronic Illnesses approved the present study. A total of 37 consecutive American patients (≥ 7 years-old) with autism spectrum disorders (ASDs) (Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition-DSM IV), born from 1983-1998, that presented to the Genetic Centers of America for outpatient genetic evaluations were prospectively examined for urinary porphyrin levels (LabCorp, Inc.) from June 2005-June 2006. Imaging and laboratory testing were conducted on each patient to rule-out other causal factors for their ASDs. As controls, age-, sex-, and race-matched neurotypical ASD siblings were examined. An apparent dose-response effect was observed between autism severity and increased urinary coproporphyrins. Patients with non-chelated autism (2.25-fold, 83% had levels > 2 SD above the control mean) and non-chelated ASDs (2-fold, 58% had levels > 2 SD above the control mean), but not patients with non-chelated pervasive developmental delay-not otherwise specified (PDD-NOS) or Asperger's disorder (1.4-fold, 46% had levels > 2 SD above the control mean), had significantly increased median coproporphyrin levels versus controls. A significant increase (1.7-fold) in median coproporphyrin levels was observed among non-chelated ASD patients versus chelated ASD patients. Porphyrins should be routinely clinically measured in ASDs, and potential ASD treatments should consider monitoring porphyrin levels. Additional research should be conducted to evaluate the potential role for mercury exposure in some ASDs.

Keywords: Autistic; Chelation; Developmental Delay

INTRODUCTION

Autism spectrum disorders (ASDs) are neurodevelopmental disorders characterized by impairments in social relatedness and communication, repetitive behaviors, abnormal movement patterns, and sensory dysfunction (Eigsti and Shapiro, 2003; Werner and Dawson, 2005). While genetic factors are recognized as being important in the pathogenesis of ASDs, a role for environmental factors has received considerable attention. Several recent epidemiological studies have associated mercury exposure with ASDs (Counter *et al.*, 2002; Holmes *et al.*, 2003; Geier and Geier, 2005; Palmer *et al.*, 2006; Windham *et al.*, in press), and it has been reported that exposure to mercury can cause immune, sensory, neurological, motor, and behavioral dysfunctions similar to traits defining or associated with ASDs, and that these similarities extend to neuroanatomy, neurotransmitters, and biochemistry (Faustman *et al.*, 2000; Bernard *et al.*, 2001; 2002; Redwood *et al.*, 2001; Blaxill *et al.*, 2004). Furthermore, a recent review has suggested that ASD children have been found to have significantly higher exposure to mercury than controls, and ASD children have been determined to have significantly increased body-burdens of mercury resulting from biochemical and genomic susceptibilities within detoxification pathways (Mutter *et al.*, 2005).

Recently, Nataf *et al.* (2006) have examined urinary porphyrin levels in a large series of children with autistic disorders from France. These researchers observed a porphyrin pattern among children with ASDs that implicated environmental toxicity, especially mercury toxicity, in childhood autistic disorders. The purpose of the present study was to conduct an examination of urinary porphyrin levels in a series of American patients with ASDs, so as to evaluate the consistency of the observations made by Nataf *et al.* in France with clinical observations made in the United States.

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