

The biological basis of autism spectrum disorders: Understanding causation and treatment by clinical geneticists

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Autism spectrum disorders (ASDs), also known as pervasive developmental disorders (PDD), are a behaviorally defined group of neurodevelopmental disorders that are usually diagnosed in early childhood. ASDs disproportionately affect male children. Mercury (Hg), a heavy metal, is widespread and persistent in the environment. Mercury is a ubiquitous source of danger in fish, drugs, fungicides/herbicides, dental fillings, thermometers, and many other products. Elevated Hg concentrations may remain in the brain from several years to decades following exposure. This is important because investigators have long recognized that Hg is a neurodevelopmental poison; it can cause problems in neuronal cell migration and division, and can ultimately cause cell degeneration and death. Case-reports of patients have described developmental regressions with ASD symptoms following fetal and/or early childhood Hg exposure, and epidemiological studies have linked exposure to Hg with an elevated risk of a patient being diagnosed with an ASD. Immune, sensory, neurological, motor, and behavioral dysfunctions similar to traits defining or associated with ASDs were reported following Hg intoxication with similarities extending to neuroanatomy, neurotransmitters, and biochemistry. The sexual dimorphism of ASDs may result from synergistic neurotoxicity caused by the interaction of testosterone and Hg; in contrast, estrogen is protective, mitigating the toxicity of Hg. Mercury exposure may significantly increase androgen levels, and as a result, patients diagnosed with an ASD may significantly benefit from anti-androgen therapy. Finally, the clinical geneticist has a wealth of biomarkers to evaluate and treat patients diagnosed with an ASD.

Key words: autistic, estradiol, ethylmercury, merthiolate, methylmercury, Thimerosal

INTRODUCTION

Autism spectrum disorders (ASDs), also known as pervasive developmental disorders (PDD), are a behaviorally defined group of neurodevelopmental disorders that are usually diagnosed in early childhood. ASDs disproportionately affect male children (roughly, 5 males per 1 female) (Austin 2008). ASDs are characterized by early onset of impairments in social interaction and communication, and the development of unusual stereotyped behaviors. Unable to learn from the natural environment as most children, the child diagnosed with an ASD generally shows little interest in the world or people around him/her. Although a few children with

an ASD develop normal and even advanced skills in particular areas, most exhibit a wide range of profound behavioral problems and delayed or undeveloped skills. Further, a child diagnosed with an ASD may display a range of problem behaviors such as hyperactivity, poor attention, impulsivity, aggression, self injury and tantrums. In addition, many frequently display unusual responses to sensory stimuli such as hypersensitivities to light or certain sounds, colors, smells, or touch and have a high threshold of pain (Austin 2008). Further, common co-morbidity conditions often associated with an ASD diagnosis include gastrointestinal disease and dysbiosis (White 2003), autoimmune disease (Sweeten et al. 2003), and mental retardation (Bolte and Poustka 2002). Therefore, in the absence of treatment, an ASD is, in general, a lifelong developmental disability that profoundly affects the way a person comprehends, communicates and relates to others.

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