

PORPHYRINURIA IN KOREAN CHILDREN WITH AUTISM: CORRELATION WITH OXIDATIVE STRESS

Seung-Il Youn¹, Soo-Hee Jin¹, Sung-Hoon Kim², Sabina Lim¹

¹Department of Basic Eastern Medical Science, Graduate School, KyungHee University, Seoul

²Cancer Preventive Material Development Research Center, College of Oriental Medicine, KyungHee University, Seoul, Republic of Korea

Autism spectrum disorder (ASD) is a neurodevelopmental disorder believed to be associated with heavy metal exposure, especially mercury (Hg), and is characterized by disturbances in metal elimination. Various studies correlated elevated heavy metal body burden with ASD diagnoses as evidenced by increased urinary porphyrin levels in patients. Urinary porphyrins were also determined in Korean patients diagnosed with ASD (n = 65) who visited AK Eastern Medicinal Clinic in Kangnam-gu, Seoul, from June 2007 to September 2008, compared to controls (n = 9) residing in the same area, by means of Metamatrix (CLIA-approved) laboratory testing. Further, urinary organic acids as indicators of hepatic detoxification/oxidative stress were also analyzed among patients diagnosed with ASD. Significant increases were found in patients diagnosed with ASD for proporphyrins, pentacarboxyporphyrin, precoproporphyrin, coproporphyrins, and total porphyrins. Significant correlations were observed between hepatic detoxification/oxidative stress markers and urinary porphyrins. In agreement with published data, the present results demonstrated that measurement of porphyrins serves as a reliable tool for diagnosis of heavy metal involvement in ASD.

Autism spectrum disorder (ASD) is a neurodevelopmental disorder characterized by impairments in social skills, cognitive ability, learning of stereotypical behaviors, bidirectional communication, overall sensory abilities, and gross and fine motor control and is manifested by abnormal behaviors (Eigsti & Shapiro, 2003; Werner & Dawson, 2005). While there is no consensus on the underlying cause of the disease, the factor of genetic inheritance is well known to play a critical role in disease development. Further, numerous epidemiological studies established a role for mercury (Hg) poisoning to be implicated in the development of ASD (Counter et al., 2002; Holmes et al., 2003; Geier & Geier, 2005, 2006a, 2006c; Geier et al., 2009c, Palmer et al., 2006, 2009; Windham et al., 2006; Young et al., 2008).

Subjects exposed to Hg poisoning experience immune dysfunction and impairments in sensory, motor, and overall neuronal system, and exhibit abnormal behavior, which resemble characteristic symptoms of patients diagnosed with ASD (Faustman et al., 2000; Bernard et al., 2001, 2002; Redwood et al., 2001; Sweet & Zelickoff, 2001; Blaxill et al., 2004; Geier et al., 2008).

Many patients diagnosed with ASD carry single-nucleotide polymorphisms in genes involved in pathways for glutathione (GSH) synthesis, resulting in reduced activity of these pathways (Buyske et al., 2006; James et al., 2006). Glutathione is critical for detoxification of Hg, and GSH was found to be significantly decreased among patients diagnosed with ASD (Geier et al., 2009a).

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Address correspondence to Sabina Lim, MD, PhD, Professor, Department of Basic Eastern Medical Science, Graduate School, KyungHee University, Seoul, Republic of Korea, #47 Gyeonghuida-gil, Dongdaemun-gu, Seoul, 130-701, Korea. E-mail: lims@khu.ac.kr