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Research Article

Severity of ASD symptoms and their correlation with the presence of copy number variations and exposure to first trimester ultrasound

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Abstract

Current research suggests that incidence and heterogeneity of autism spectrum disorder (ASD) symptoms may arise through a variety of exogenous and/or endogenous factors. While subject to routine clinical practice and generally considered safe, there exists speculation, though no human data, that diagnostic ultrasound may also contribute to ASD severity, supported by experimental evidence that exposure to ultrasound early in gestation could perturb brain development and alter behavior. Here we explored a modified triple hit hypothesis [Williams & Casanova, 2010] to assay for a possible relationship between the severity of ASD symptoms and (1) ultrasound exposure (2) during the first trimester of pregnancy in fetuses with a (3) genetic predisposition to ASD. We did so using retrospective analysis of data from the SSC (Simon's Simplex Collection) autism genetic repository funded by the Simons Foundation Autism Research Initiative. We found that male children with ASD, copy number variations (CNVs), and exposure to first trimester ultrasound had significantly decreased non-verbal IQ and increased repetitive behaviors relative to male children with ASD, with CNVs, and no ultrasound. These data suggest that heterogeneity in ASD symptoms may result, at least in part, from exposure to diagnostic ultrasound during early prenatal development of children with specific genetic vulnerabilities. These results also add weight to on-going concerns expressed by the FDA about non-medical use of diagnostic ultrasound during pregnancy. Autism Res 2016. © 2016 International Society for Autism Research, Wiley Periodicals, Inc.

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