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Birth Defects In Children With Autism Spectrum Disorder And Co-occurring Intellectual Disability

What is this research about?

Birth defects form between conception and birth. Especially during the 3rd to 8th week of pregnancy (when major organs are formed), both genetic and environmental factors influence growth and may result in birth defects and abnormal brain development. There are more defects in the central nervous system (CNS) reported in people with ASD. Also, ASD is linked to head and facial bone differences such as large head circumference, high palate, cleft palate and cleft lip, and abnormal distance between eyes. This study examined the relationship between individuals with ASD, with and without ID, and birth defects in different organ systems (such as the CNS, eye, ear, face and neck, cardiovascular, respiratory, clefts, gastrointestinal, genitourinary, musculoskeletal, skin).

What did the researchers do?

Researchers conducted a case-control study based on data collected from numerous nationwide registries of children born in Finland between 1987 and 2007. Control participants (no ASD or ID; n = 17 727) were matched with 4441 children diagnosed with ASD in a 4:1 ratio based upon date of birth, sex, place of birth and residence. Researchers analyzed the relationship between having ASD and birth defects, as well as birth weight, prematurity, weight for gestation age, maternal hemorrhage, birth injury or trauma, and multiple births.

What you need to know:

There is a higher chance of various birth defects occurring during early pregnancy, in children with autism spectrum disorder (ASD). Children with both ASD and intellectual disability (ID) are more likely to have birth defects compared to children who have ASD without ID.

What did the researchers find?

Children with ASD were twice as likely to have single and multiple birth defects and congenital syndromes compared to controls. Birth defects related to the CNS (i.e. spina bifida - incomplete closing of the spine and spinal cord) were 30 times more likely among ASD children compared to controls, and encephalocele (sac-like growths of the brain and its membranes) were also more common. In addition, having both ASD and birth defects was linked to low birth weight (under 2.5 kg) and prematurity (under 37 weeks). Lastly, a stronger association existed between people with ASD and ID and organ system birth defects compared to those with ASD without ID. The strongest associations for those with both ASD and ID were observed for birth defects of the eye (5x more likely), face and neck (9x), and CNS (12x), orofacial clefts (7x) and the musculoskeletal system (5x). Overall, these findings suggest that the causes of some forms of ASD predate organ formation and are present in the womb.

How can you use this research?

This research helps with identifying and classifying prenatal risk factors of ASD, such as low birth weight, being small for gestational age, maternal hemorrhage, birth injury or trauma, and multiple births. Also, understanding the timeline of events during the first trimester of pregnancy will advance research of genetic and environmental interactions that influence brain development.

About the Researchers

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About the Chair

The Chair in Autism Spectrum Disorders Treatment and Care Research is dedicated to studying ways to improve the mental health and well-being of people with Autism Spectrum Disorders (ASD) and their families in Canada.

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For more information, visit the Chair in Autism Spectrum Disorders Treatment and Care Research website at asdmentalhealth.ca

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