Improving Autism Diagnosis

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Autism is a pervasive developmental disorder with a wide range of symptoms that is increasing in occurrence throughout the population. It typically becomes evident in early childhood, with indications including lack of social skills, delayed communication, echolalia, and repetitive behaviors, among many others. Because of the broad range in symptoms, and their "gradual and inconsistent" onset, it has been difficult to create definitive screening techniques to determine possible genetic and/or environmental risk factors that contribute to the manifestation of autism. What makes one susceptible?

The most definitive symptoms of the autism spectrum disorder umbrella are social development abnormalities, which, scientifically speaking can be difficult to test young children for because their social evolution does not follow a predictable timetable. They each reach milestones at different ages. When conducting studies, scientists must also be cautious so as not to cross ethical boundaries. Testing effectively is challenging and more prone to error when it is limited (ex: no control group, based solely on behavior).

A new approach for autism screening has been created for use during the 1 year well child check-up. It involves "…examining early markers related to early brain overgrowth, cerebellar development, gene expression patterns and immune system (dys)function."[1] Evidence indicates that something has occurred within in the first year of life that leads to development of this

[1] P. 132

disorder. By measurement of head circumference it has been discovered that babies who are later diagnosed with autism experience rapid brain overgrowth during the first year, followed by an immediate arrest of growth. This coincides with the "plummet in social behavior". Synthesis of brain serotonin has been found to be abnormal. T-lymphocyte (immunity and infection fighters) number and function is atypical. A defect exists in the pleiotropic MET (mesenchymal epithelial transition factor) gene, which can affect cerebellar growth and immune system function. Variations found in genes UBE3A (ubiquitin protein ligase E3A), GABRB3 (gamma aminobutyric acid A receptor, beta 3), ASS1 (argininosuccinate synthase 1) and NAGLU (N-acetylglucosaminidase, alpha),among others, have been shown to be associated with differences in the cell cycle, signal transduction, metabolism and development. Discovery of these brain and blood biomarkers are indicative of the promising possibilities that lie ahead regarding prevention, diagnosis and treatment of autism.

Naturally the hope is to one day have a specific, reliable screening mechanism in place. Diagnosis of autism is devastating, difficult to accept, and even more difficult to understand. "Detecting autism at the earliest possible age is of the utmost importance to optimize outcomes for children with the disorder."[2]

[2] P. 133

Bibliography

1. https://www.aacp.com/pdf%2F2103%2F2103ACP_Review1.pdf

2. http://www.genecards.org/cgi-bin/carddisp.pl?gene=MET

3. http://ghr.nlm.nih.gov/gene

As the mother of a child with autism, I hope that more discoveries continue to be made, and sooner rather than later. There are no words that can describe the emotion you feel when you find something could be wrong with your child. Having read over the years of possible causes for my son's autism, reading this article provided a little comfort. Hearing his diagnosis was difficult to accept, because you always want nothing but the best for your child, health being at the top of the list. While it can sometimes be hard for us to understand his limitations, I can't imagine what it is like for him, and it makes me sad that he can't always tell me what he is feeling. But it's all a learning process. We all agree he is the most wonderful boy in the world, and is extremely affectionate and sweet, and we are so lucky to have him in our lives. I am thankful that Jack Later let me write my paper on this article.