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Genes and Autism

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We are often asked about the genetics of autism and recent research findings. This is a complex subject. I have provided some websites addresses that are well written and give fairly straight forward information. There are also references to recent scientific papers (published within the past 12 months). Abstracts for these appear below.

Useful Websites

The Exploring Autism website is the result of collaboration between researchers, non-profit groups, and families who are living with autism. Organizations that make this site possible range from major universities and medical centers to the National Alliance for Autism Research.

Easy to read information about the genetics of autism and recent research.

www.exploringautism.org

Medline Plus. A health information service of the US National Library of Medicine and the National Institutes of health.

The link below takes you to a summary of genes and autism research. It includes a very useful glossary of terms and a simple introduction “What are genes?”

www.nichd.nih.gov/publications/pubs/upload/autism_genes_2005.pdf

The National Autistic Society UK champions the rights and interests of all people with autism and aim to provide individuals with autism and their families with help, support and services that they can access, trust and rely upon and which can make a positive difference to their lives. The website includes information about autism and Asperger syndrome, the NAS and its services and activities.

Follow the link below to a briefing on genetics and autism.

www.nas.org.uk/nas/jsp/polopoly.jsp?d=115&a=3578

Recent Papers

1. **Abha R. Gupta and Matthew W. State. (2007). Recent Advances in the Genetics of Autism.** ***Biol Psychiatry, 61: 429-437***

   Abstract: Autism is a strongly genetic disorder, with an estimated heritability of greater than 90%. Nonetheless, its specific genetic etiology remains largely unknown. Over the past several years, the convergence of rapidly advancing genomic technologies, the completion of the human genome project, and successful collaborative efforts to increase the number of deoxyribonucleic acid samples available for study have led to the first solid clues regarding the genetic origins of autism spectrum disorders. This article addresses the obstacles that have confronted gene discovery efforts and reviews recent linkage, cytogenetic, and candidate gene association studies relevant to autism spectrum disorders. In addition, promising avenues for future research and the potential contribution of emerging genomic technologies are considered.

2. **Mao Sheng Yang and Michael Gill (2007). A review of gene linkage, association and expression studies in autism and an assessment of convergent evidence.** ***Int. J. Devl Neuroscience 25: 69-85***

   Abstract: Autism is a neurodevelopmental disorder with high heritability and a likely complex genetic architecture. Much genetic evidence has accumulated in the last 20 years but no gene has been unequivocally identified as containing risk variants for autism. In this article we review the past and present literature on neuro-pathological, genetic linkage, genetic association, and gene expression studies in this disorder. We sought convergent evidence to support particular genes or chromosomal regions that might be likely to contain risk DNA variants. The convergent evidence from these studies supports the current hypotheses that there are multiple genetic loci predisposing to autism, and that genes involved in neurodevelopment are especially important for future genetic studies. Convergent evidence suggests the chromosome regions 7q21.2–q36.2, 16p12.1–p13.3, 6q14.3–q23.2, 2q24.1–q33.1, 17q11.1–q21.2, 1q21–q44 and 3q21.3–q29, are likely to contain risk genes for autism. Taken together with results from neuro-pathological studies, genes involved in brain development located at the above regions should be prioritized for future genetic research.

3. **Robert Delong (2007). GABA(A) receptor alpha5 subunit as a candidate gene for autism and bipolar disorder: A proposed endophenotype with parent-of-origin and gain-of-function features,with or without oculocutaneous albinism.** ***Autism, Vol. 11, No. 2, 135-147***

   Abstract: Our earlier family history studies of individuals with autism found a high incidence of major affective disorder, especially bipolar disorder, and unusual talents or intellectual abilities among family members. We now describe a subgroup of such families, selected from a large clinical experience, illustrating specific features of major affective disorder, special talents or intellectual ability, and familial patterns of trait transmission, with the additional feature of oculocutaneous albinism in some cases. These observations, suggesting parent-of-origin and gain-of-function effects, considered together with recent genetic findings in the literature, suggest a genetic hypothesis possibly unifying disparate observations found in families of individuals with autism.