Director's Blog
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NIMH's Top 10 Research Advances of 2011

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At NIMH and in our broad research community, this has been a year of exciting discoveries and scientific progress, as we strive to make a difference for those with mental illness. Here are 10 breakthroughs and events of 2011 that are changing the landscape of mental health research.

4. De Novo Genetic Variants.

This year scientists looking at families with only one case of autism found that up to eight percent of cases in these families were the result of de novo (unique to the person affected) copy-number variants—stretches of DNA that were either multiplied or truncated (9, 10). Analysis of the gene regions affected by these variants implicated a network of genes involved in the development of synapses and neuronal function (11). **Another study, focusing specifically on sequences of DNA that code for protein, yielded other de novo genetic changes in one-case families (12). While providing information on genetic contributors to a significant fraction of sporadic autism cases, the work also reveals gene regions for future investigation and ultimately, information on functional changes underlying autism that will offer clues to therapy.**

References

(12) **O'Roak BJ**, Deriziotis P, Lee C, et al. Exome sequencing in sporadic autism spectrum disorders identified severe de novo mutations. *Nat Genet.* 2011 Jun;43(6):585-9.