

SPECIAL REPORT SUBARTICLE

Notable papers of 2014

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It's no easy feat to whittle down the list of the most influential autism papers to a mere 10. So please consider this but a taste of the burgeoning field, presented in chronological order and based on suggestions from many researchers. SFARI director **Louis Reichardt** elaborates on the list in his **year-end column**, noting that it is "dominated by genetic studies" that will transform autism research.

1. Spontaneous and rare mutations are key in schizophrenia

Guest blog: Schizophrenia milestone holds lessons for autism

Three massive sequencing studies in the field of schizophrenia offer clues about autism and highlight the need for large study samples when searching for rare mutations and common variants.

Fromer M. *et al. Nature* **506**, 179-184 (2014) [PubMed](#)

Purcell S.M. *et al. Nature* **506**, 185-190 (2014) [PubMed](#)

Schizophrenia Working Group of the Psychiatric Genomics Consortium. *Nature* **511**, 421-427 (2014) [PubMed](#)

2. Girls protected from autism, study suggests

Autism characteristics differ by gender, studies find

A string of studies published earlier this year suggest that the causes and manifestations of autism differ in girls and boys.

Jacquemont S. *et al. Am. J. Hum. Genet.* **94**, 415-425 (2014) [PubMed](#)

Frazier T.W. *et al. J. Am. Acad. Child Adolesc. Psychiatry* **53**, 329-340 (2014) [PubMed](#)

3. **Odd brain patches point to origins of autism in womb**

Small swaths of what look like immature neurons bespeckle the brains of some children with autism, according to a study of postmortem brains.

Stoner R. *et al. N. Engl. J. Med.* **370**, 1209-1219 (2014) [PubMed](#)

4. **List of gene 'hot spots' is rich with autism mutations**

Autism-linked mutations are six times more likely to fall in genetic regions that are critical for brain function than elsewhere in the genome.

Uddin M. *et al. Nat. Genet.* **46**,742-747 (2014) [PubMed](#)

5. **Cluster of symptoms reveals gene's link to autism subtype**

A gene that regulates the structure of DNA, called CHD8, may be the closest thing to an autism gene so far.

Bernier R. *et al. Cell* **158**, 263-276 (2014) [PubMed](#)

6. **Common mutations account for half of autism risk**

Mutations that are present in 5 percent or more of the population may add up to trigger autism.

Gaugler T. *et al. Nat. Genet.* **46**, 881-885 (2014) [PubMed](#)

7. **Brains of children with autism teem with surplus synapses**

The pruning process that eliminates unnecessary brain connections in the first years of life may go awry in autism.

Tang G. *et al. Neuron* **83**, 1131-1143 (2014) [PubMed](#)

8. [New therapy shows promise for infants with signs of autism](#)

The interaction-based therapy, which is delivered by parents at home, improved behavior and language skills in a small pilot study.

Rogers S.J. *et al. J. Autism Dev. Disord.* **44**,2981-2995 (2014) [PubMed](#)

9. **Large study links maternal infection to autism risk**

Having a serious infection during pregnancy raises the risk of having a child with autism by 37 percent, according to a study of more than 2.3 million people.

Lee B.K. *et al. Brain Behav. Immun.* Epub ahead of print (2014) [PubMed](#)

10. **Massive sequencing studies reveal key autism genes**

A mammoth effort to analyze the sequences of more than 20,000 people unearthed the longest and most robust list of autism genes yet.

Iossifov I. *et al. Nature***515**, 216-221 (2014) [PubMed](#)

De Rubeis S. *et al. Nature***515**, 209-215 (2014) [PubMed](#)