

SPECIAL REPORT

Notable papers of 2011

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The past year saw the publication of many groundbreaking papers on autism, making our task of picking just ten a particularly arduous one. Below is a chronological list of just some of the papers, selected by SFARI staff. **In his essay**, SFARI scientific director Gerald Fischbach describes his thoughts on the list.

1. **Study finds high rate of autism in South Korea**

The first comprehensive autism study in South Korea found in May that the **prevalence** of the disorder is more than double the number in the United States.

Kim Y.S. *et al. Am. J. Psychiatry* **168**, 904-912 (2011) [PubMed](#)

2. **Family sequencing study boosts two-hit model of autism**

Sequencing the entire **exome** — the protein-coding parts of the genome — of 20 children with autism and their unaffected parents, researchers in May reported four *de novo*, or spontaneous, mutations in autism-associated genes.

O'Roak B.J. *et al. Nat. Genet.* **43**, 585-589 (2011) [PubMed](#)

3. **Networks of genes altered in autism brains, study says**

A May study of gene expression in postmortem brains found that two networks of genes are abnormally expressed in the brains of people with autism.

Voineagu I. *et al. Nature* **474**, 380-384 (2011) [PubMed](#)

4. Studies find high rate of rare new mutations in autism

In June, three new studies analyzing genetic data from families in which just one child has autism reported the strongest evidence yet that rare new mutations contribute to the disorder.

Sanders S.J. *et al. Neuron* **70**, 863-885 (2011) [PubMed](#)

Levy D. *et al. Neuron* **70**, 886-897 (2011) [PubMed](#)

Gilman S.R. *et al. Neuron* **70**, 898-907 (2011) [PubMed](#)

5. Protein networks link different forms of autism, study says

Also in June, researchers identified hundreds of previously unknown connections between proteins involved in autism spectrum disorders.

Sakai Y. *et al. Sci. Transl. Med.* **3**, 86ra49 (2011) [PubMed](#)

6. Experts critique statistics, conclusion of autism twin study

A new study of twins, published in July, proposed the controversial claim that environmental influences during early development are just as important as genetics, if not more so.

Hallmayer J. *et al. Arch. Gen. Psychiatry* **68**, 1095-1102 (2011) [PubMed](#)

7. Large study finds 'baby sibs' at high risk of autism

Children who have more than one older sibling with autism have a one in three chance of developing the disorder themselves, according to a study published in August.

Ozonoff S. *et al. Pediatrics* Epub ahead of print (2011) [PubMed](#)

8. Autism tests struggle to balance accuracy and speed

Whether someone is diagnosed with classic autism, **Asperger syndrome** or pervasive developmental disorder—not otherwise specified varies significantly among different clinical sites, even when they are all relying on the same diagnostic tests, according to a study published in

November.

Lord C. *et al. Arch. Gen. Psychiatry* Epub ahead of print (2011) [PubMed](#)

Tuberous sclerosis, fragile X may be molecular opposites

Some forms of autism are caused by too many proteins at the junction between neurons, whereas other forms result from too few, suggesting that drugs for one form of autism may not help, and may even harm, individuals with another, according to a report in November.

Auerbach B.D. *et al. Nature* **480**, 63-68 (2011) [PubMed](#)

Neurons made from stem cells reveal cellular flaws in autism

In December, researchers reported that neurons regenerated from individuals with Timothy syndrome, a rare autism-related disorder, have several cellular abnormalities.

Pa?ca S.P. *et al. Nat. Med.* **17**, 1657-1662 (2011) [PubMed](#)