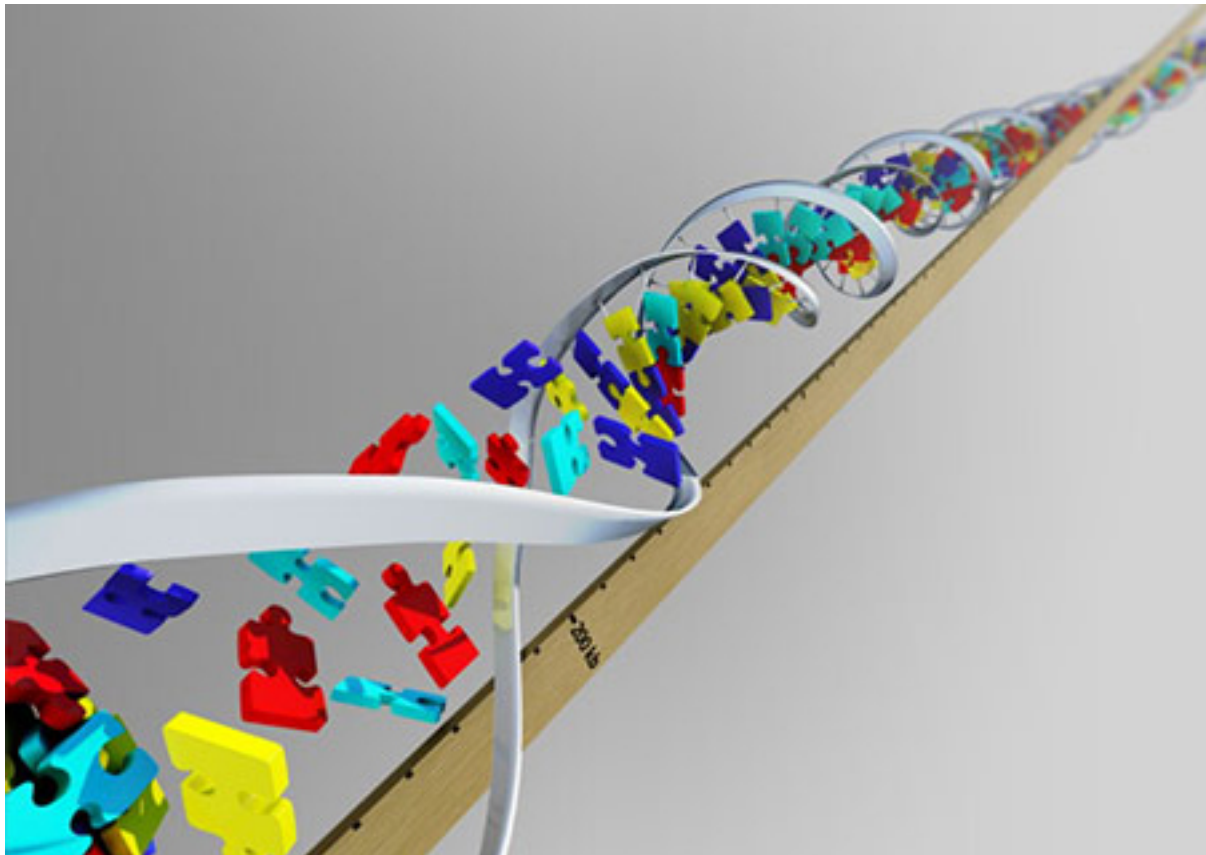


SPECIAL REPORT SUBARTICLE

Notable papers of 2013

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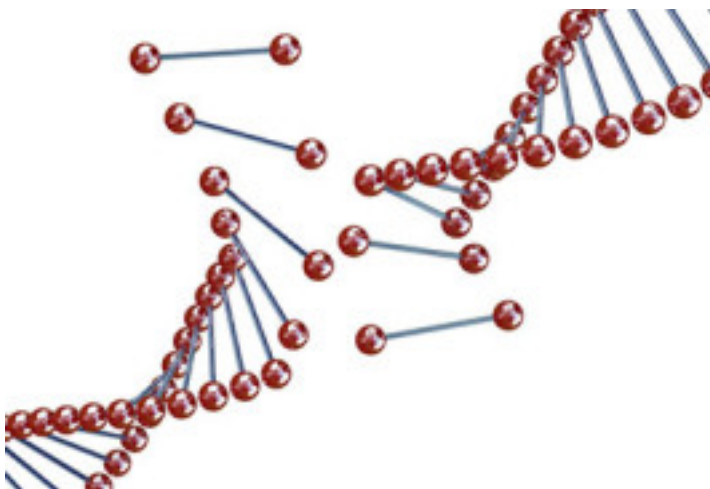
Our annual list of the most influential papers in the autism field is culled from suggestions solicited from SFARI staff, as well as from experts in the field. Below is a selection of just some of these papers, presented in chronological order. SFARI director Louis Reichardt expounds on the list in [this column](#).



1. **Can nutritional supplements help treat some cases of autism?**

A handful of studies published in late 2012 and early 2013 point to dietary deficiencies as a contributing factor in some forms of autism, suggesting that supplements such as folic acid may help treat and even prevent the disorder.

Surén P. *et al.* *JAMA* **309**, 570-577 (2013) [PubMed](#)



2. **Genome-editing tools compose new models of autism**

Thanks to a suite of new tools based on synthetic biology, it's now possible to quickly and cheaply insert autism-linked mutations into living cells in the lab, according to two studies published 15

February in *Science*.

Cong L. *et al. Science* **339**, 819-823 (2013) [PubMed](#)

Mali P. *et al. Science* **339**, 823-826 (2013) [PubMed](#)



3. [Twin study suggests girls are protected from autism risk](#)

A comparison of autism-like behaviors in nearly 10,000 pairs of fraternal twins suggests that girls are somehow protected from the disorder. The findings, published 19 February in the *Proceedings of the National Academy of Sciences*, may partly explain why autism is four times more common in boys than in girls — one of the oldest and most puzzling statistics in the field.

Robinson E.B. *et al. Proc. Natl. Acad. Sci. USA* **110**, 5258-5262 (2013) [PubMed](#)



4. **Statins improve symptoms of Rett syndrome in mice**

Defects in cholesterol metabolism may influence the severity of Rett syndrome, suggesting a treatment for the autism-related disorder, according to research published in July in *Nature Genetics*. We first reported these findings from the 2013 **International Meeting for Autism Research** in May.

Buchovecky C.M. *et al. Nat. Genet.* **45**, 1013-1020 (2013) **PubMed**

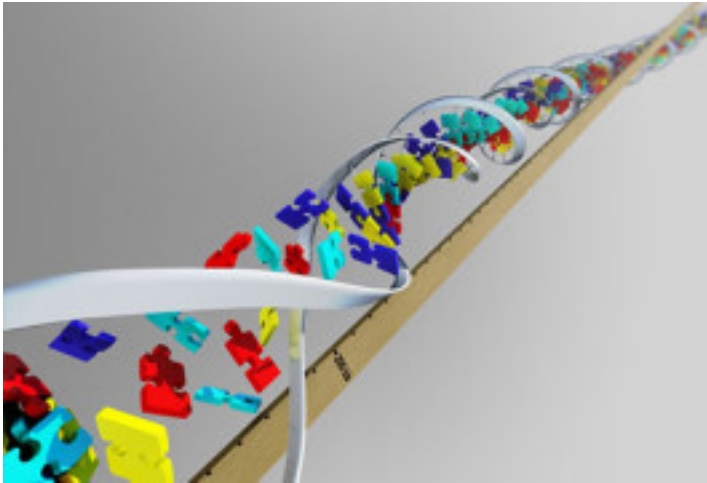


5. **Gene therapy reverses Rett syndrome symptoms in mice**

A virus that ferries healthy copies of the Rett syndrome gene across the blood-brain barrier can reverse symptoms in female mice that model the disorder, according to a report published 21

August in the *Journal of Neuroscience*.

Garg S.K. *et al. J. Neurosci.* **33**,13612-13620 (2012) [PubMed](#)



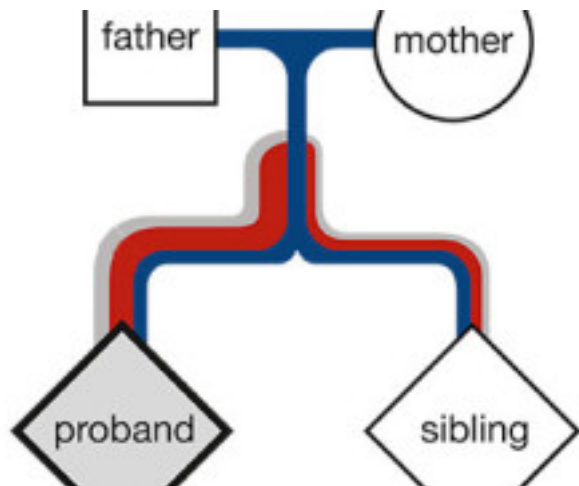
6. [Autism genes are surprisingly large, study finds](#)

[Length matters: Disease implications for long genes](#)

[What does the existence of long genes tell us?](#)

Enzymes called topoisomerases are crucial for the expression of extremely long genes in neurons, according to a study published 5 September in *Nature*. More than one-quarter of these genes are known autism candidates, the study found. The paper set off a flurry of discussion, including a [Viewpoint](#) by the lead researcher [Mark Zylka](#) about the implications of this finding for autism.

King I.F. *et al. Nature* **501**, 58-62 (2013) [PubMed](#)

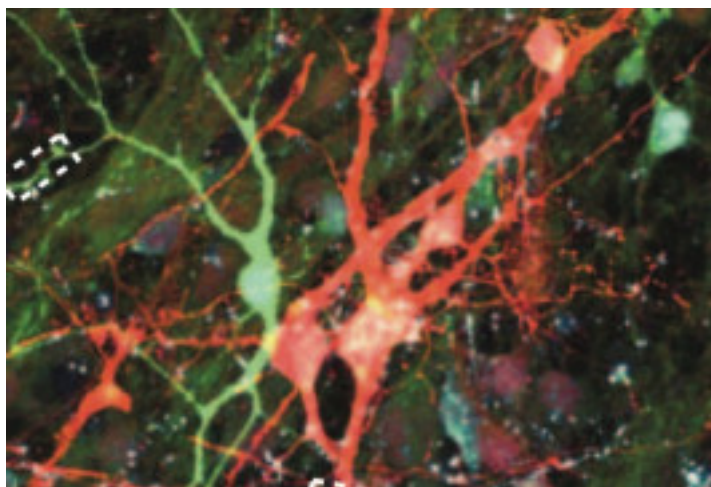


7. **Small deletions, duplications of DNA may up autism risk**

Two new studies, published in September and October, found more small deletions and duplications of DNA in individuals with autism than in controls. These variants may also affect the severity of the disorder.

Krumm N. *et al. Am. J. Hum. Genet.* **93**, 595-606 (2013) [PubMed](#)

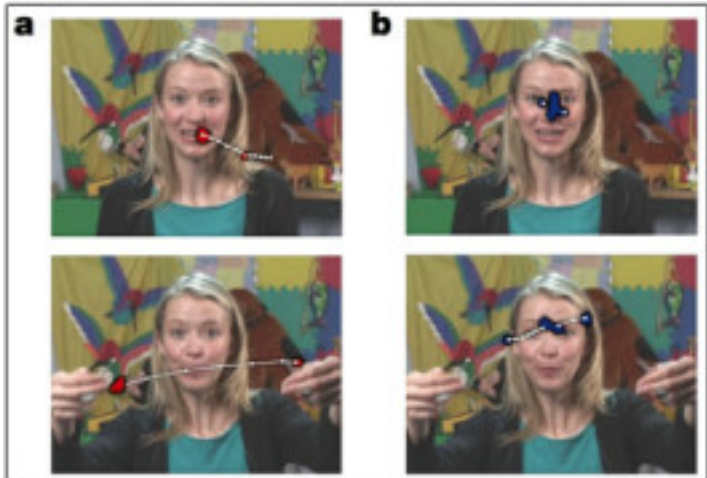
Poultney C.S. *et al. Am. J. Hum. Genet.* **93**, 607-619 (2013) [PubMed](#)



8. **Drug fixes cellular defects in autism-related disorder**

A new stem-cell model of Phelan-McDermid syndrome points to a possible treatment for the rare autism-related disorder, according to a study published 16 October in *Nature*.

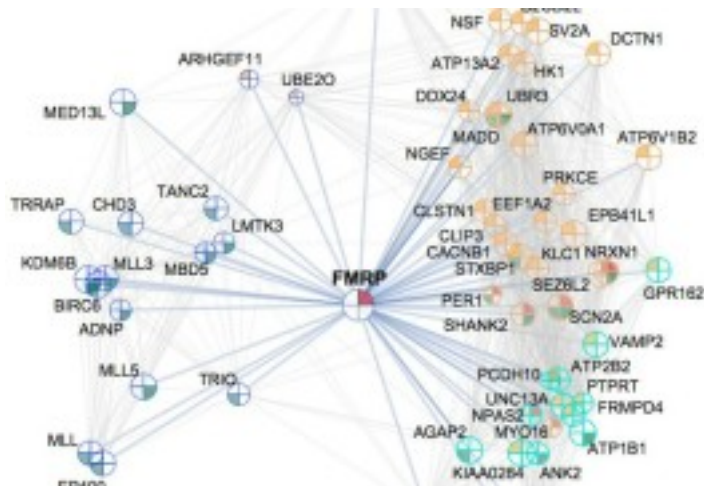
Shcheglovitov A. *et al. Nature* **503**, 267-271 (2013) [PubMed](#)



9. **Classic sign of autism appears early in infancy, study says**

Baby boys who will later be diagnosed with autism show a loss of interest in other people's eyes between 2 and 6 months of age, according to a study published 6 November in *Nature*. This is the earliest behavioral marker of autism found to date.

Jones W. and A. Klin *Nature* Epub ahead of print (2013) [PubMed](#)



10. **Studies map gene expression across brain development**

Now that genetic studies have implicated several hundred genes in autism, researchers are turning their attention to where and when in the healthy young brain these genes are expressed. The first two studies to tackle these questions appeared 21 November in *Cell*.

Willsey A.J. *et al. Cell* **155**, 997-1007 (2013) [PubMed](#)

Parikshak N.N. *et al. Cell* **155**, 1008-1021 (2013) [PubMed](#)