

**SPECIAL REPORT SUBARTICLE**

# Notable papers of 2012

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Picking the top ten papers in a field is always challenging, but the outpouring of high-quality autism research in 2012 made that task especially difficult. Below is a selection of just some of these papers, selected by SFARI staff and presented in chronological order. SFARI director Gerald Fischbach expounds on the list in [this column](#).

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## 1. **Brain response to gaze predicts autism in baby sibs**

A longitudinal study of **infant siblings** of children with autism is the first to identify a **particular brain pattern** that is linked to later diagnosis of autism.

Elsabbagh M. *et al. Curr. Biol.* 22, 338-342 (2012) [PubMed](#)



## 2. **Bone marrow transplant curbs symptoms of autism and Rett syndrome**

Two papers found that a bone marrow transplant can alleviate autism-like symptoms in mice, one

in a **rodent model of Rett syndrome** and the other in an **immune model of autism**.

Derecki N.C. *et al. Nature* **484**, 105-109 (2012) [PubMed](#)

Hsiao E.Y. *et al. Proc. Natl. Acad. Sci. USA* **109**, 12776-12781 (2012) [PubMed](#)



### 3. **Hundreds of genes involved in autism, sequencing studies say**

A quartet of **exome-sequencing studies** published this year have identified new risk genes and **point to FMRP**, the protein implicated in **fragile X syndrome**, as a central hub.

O’Roak B.J. *et al. Nature* **485**, 246-250 (2012) [PubMed](#)

Sanders S.J. *et al. Nature* **485**, 237-241 (2012) [PubMed](#)

Neale B.M. *et al. Nature* **485**, 242-245 (2012) [PubMed](#)

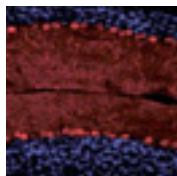
Iossifov I. *et al. Neuron* **74**, 285-299 (2012) [PubMed](#)



### 4. **Chromosome exchanges reveal new autism-related genes**

**Large-scale swapping of genetic material** between chromosomes may play an important role in autism, according to a study published 27 April in *Cell*.

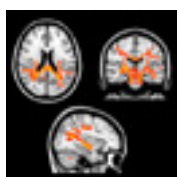
Talkowski M.E. *et al. Cell* **149**, 525-537 (2012) [PubMed](#)



### 5. **Tuberous sclerosis gene loss triggers autism-like features**

Losing one or both copies of TSC1, one of the two genes responsible for tuberous sclerosis complex, in **specific cells of the cerebellum** can trigger several autism-like behaviors in mice, according to research published 1 July in *Nature*.

Tsai P.T. *et al. Nature*. **488**, 647-651 (2012) [PubMed](#)



### 6. **Autism risk gene found to alter brain wiring**

MET, a leading candidate gene for autism risk, **influences the strength of connections** between brain regions involved in social behaviors, and this effect is especially prominent in people with the disorder. The findings are from a large study using several imaging techniques, published 6 September in *Neuron*.

Rudie J.D. *et al. Neuron* **75**, 904-915 (2012) [PubMed](#)

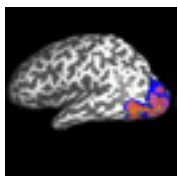


### 7. **Drug improves social deficits in fragile X syndrome**

A drug called arbaclofen **improves behavioral problems** in people with fragile X syndrome, an inherited condition that can lead to mental retardation and autism, according to the results of a clinical trial published 19 September in *Science Translational Medicine*. A second study published in the same issue showed that the drug restores normal brain function in a mouse model of the syndrome.

Berry-Kravis E.M. *et al. Sci. Transl. Med.* **4**, 152ra127(2012) [PubMed](#)

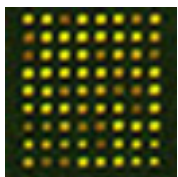
Henderson C. *et al. Sci. Transl. Med.* **4**, 152ra128 (2012) [PubMed](#)



#### 8. **'Noisy' brain signals could underlie autism, study says**

Sensory responses in the brain of an individual with autism **vary much more** than in someone without the disorder, according to a study published 20 September in *Neuron*. This may explain why some people with autism are extremely sensitive to lights and sounds.

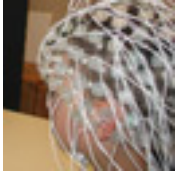
Dinstein I. *et al. Neuron* **75**, 981-991 (2012) [Abstract](#)



#### 9. **Common variants, en masse, may add up to strong autism risk**

Individually, common genetic variants confer little risk for autism. But taken together, **they may contribute significantly**, predicts a statistical analysis published 15 October in *Molecular Autism*.

Klei L. *et al. Mol. Autism* **3**, 9 (2012) [PubMed](#)



10. **Behavioral treatment for autism may normalize brain activity**

Early intensive therapy **may normalize the brain's response** to faces in young children with autism, according to a study published in the November issue of the Journal of the American Academy of Child and Adolescent Psychiatry. The results are part of a randomized, controlled trial of a treatment called the Early Start Denver Model.

Dawson G. *et al.* *J. Am. Acad. Child Adolesc. Psychiatry* **51**, 1150-1159 (2012) [PubMed](#)