

# Scientists Cure Fragile X in Mice

By Emily Singer, [www.technologyreview.com](http://www.technologyreview.com)

*A similar approach might soon be tested in humans.*

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Tweaking the production of a single gene can relieve most symptoms of Fragile X syndrome--the most common inherited cause of mental retardation and autism--in a mouse model of the disease.

Fragile X is linked to mutations in a gene on the X chromosome called FMR1. One theory of the disease is that mutations in this gene lead to over expression of a specific receptor in the brain. So Mark Bear, a neuroscientist at **Massachusetts Institute of Technology** who led the work, and his colleagues engineered mice with a similar mutation in FMR1 to produce less of that receptor. The engineered animals suffered fewer seizures--a hallmark of Fragile X--and had a normal brain structure compared with their diseased counterparts.

While it's not yet clear if the findings will translate to humans, scientists hope to learn the answer quickly. Drugs that inhibit this receptor already exist, and Bear has formed a company, Seaside Therapeutics, in Cambridge, MA, to test these drugs in clinical trials.

According to an article at Nature News,

Seaside Therapeutics has applied to the US Food and Drug Administration to test a compound, one of several mGluR antagonists licensed from Merck, in adult humans. If approved, drug trials could start as early as next year--but this class of drugs has traditionally produced mixed results and no compounds are currently approved for any condition.

Bear says that the findings might also apply to autism:

Only a small percentage of people with autism have the fragile-X mutation, but many people with fragile X satisfy the diagnostic criteria for autism. "I'd be extremely pleased if all we accomplished was correcting fragile X in humans," says Bear. "But we think and it's possible that this work on fragile X could extend into autism in general."

Still, some questions remain. Mark Hirst, scientific advisor to the United Kingdom's Fragile X Society, told the BBC,

"Whilst we know that many proteins are regulated by the fragile X protein, and are therefore disrupted in fragile X individuals, mGluR5 seems to be one of the most important."

However, he stressed that the mice in the study had benefited from reduced levels of mGluR5 throughout their development--something it would ... not be able to replicate in a human drug treatment.

He added: "We must not take our eye off the other proteins that are mis-regulated, as the basis of fragile X syndrome is likely to be more complex and involve other pathways."

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