Fragile X study offers new drug hope

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(Medical Xpress)—An experimental drug can improve sociability in patients with fragile X syndrome and may be helpful as a treatment for autism, according to a study.

Fragile X is a rare genetic disorder that affects about 1 in 4,000 boys and 1 in 8,000 girls.

It usually results in intellectual impairment and—in many cases—some form of autism.

Brain disorder

In fragile X, a mutation in a gene on the X chromosome turns off production of a regulatory protein known as FMRP.

This leads to out-of-control activation of the brain chemical glutamate, which plays a key role in learning and memory.

This could help to explain social anxiety and other symptoms of the disorder.

Reducing symptoms

Researchers at the University's Patrick Wild Centre for Autism, Fragile X Syndrome and Intellectual Disabilities tested a drug known as STX209 in mice that were genetically engineered to have a form of Fragile X.

The team found that it helped correct the biochemical abnormalities associated with the mutation.

This, in turn, reduced seizures and repetitive behaviours in the mice.

The paper has been published in *Science Translational Medicine*.

"Our paper shows that many of the changes in brain cells that are believed to underlie fragile x syndrome can be reversed by this drug. Significant advances are being made in this field and we are hopeful that on-going medical research will make a real difference to the lives of individuals with these conditions. The results are exciting because they show that a drug could help to improve social behaviour in people with fragile x," says Prof Peter Kind, co-director of the Patrick Wild Centre.

In a related study, 46 children and 17 adults with Fragile X were assigned to take the drug for four weeks and a placebo for four weeks.

Patients made bigger improvements on a "social avoidance" scale while they were taking the drug compared with when they were taking the placebo.

The research was conducted by Rush University, the UC Davis MIND Institute and Seaside Therapeutics.

Provided by University of Edinburgh