

Fragile X study offers hope of new autism treatment

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Quinn, an autistic boy, and the line of toys he made before falling asleep. Repeatedly stacking or lining up objects is a behavior commonly associated with autism. Credit: Wikipedia.

People affected by a common inherited form of autism could be helped by a drug that is being tested as a treatment for cancer.

Researchers who have identified a chemical pathway that goes awry in the brains of Fragile X patients say the drug could reverse their behavioural symptoms.

The scientists have found that a known naturally occurring chemical called cercosporamide can block the pathway and improve sociability in mice with the condition.

The team at the University of Edinburgh and McGill University in Canada identified a key molecule - eIF4E - that drives excess protein production in the brains of Fragile X patients.

This can cause behavioural symptoms that include

learning difficulties. It can also lead to more serious [intellectual disabilities](#), delays in speech and language development and problems with social interactions.

They found that eIF4E regulates the production of an enzyme called MMP-9, which breaks down and re-orders the connections between [brain cells](#) called synapses. This disrupts communication between brain cells leading to changes in behaviour.

Treatment with cercosporamide blocks the activity of eIF4E, and therefore reduces the amounts of MMP-9, and reverses the behavioural symptoms in mice with a version of Fragile X Syndrome, the team showed.

Cercosporamide is being tested as a treatment for lung cancer and [acute myeloid leukemia](#). The new findings suggest that it could also have a use as a treatment for patients with Fragile X Syndrome.

Fragile X Syndrome is the most common genetic cause of [autism spectrum disorders](#). It affects around 1 in 4000 boys and 1 in 6000 girls. Currently, there is no cure.

The study is published in the journal *Cell Reports*.

Dr Christos Gkogkas, a Chancellor's Fellow at the University of Edinburgh's Patrick Wild Centre for Research into Autism, Fragile X Syndrome and Intellectual Disabilities, said: "Our findings open the door to targeted treatments for Fragile X Syndrome. By designing treatments that block just this pathway, it is hoped that we can limit the potential side-effects and develop therapies that are more efficient than general treatment approaches."

Professor Peter Kind, Director of the Patrick Wild Centre at the University of Edinburgh, said: "Dr. Gkogkas is an important addition to the Patrick Wild Centre. His research provides important insights

into the brain processes underlying the symptoms of people with Fragile X Syndrome and other intellectual disabilities. Only with this knowledge can better medicines to treat these conditions be developed."

Professor Nahum Sonenberg, of McGill University, said: "We found that eIF4E regulates the production of an enzyme called MMP-9, which breaks down and re-orders the connections between brain cells called synapses. Excess MMP-9 disrupts communication between brain cells, leading to changes in behaviour."

More information: C. G. Gkogkas et al. Pharmacogenetic Inhibition of eIF4E-Dependent Mmp9 mRNA Translation Reverses Fragile X Syndrome-like Phenotypes. *Cell Reports*, 27 November 2014.

Provided by University of Edinburgh

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