Brain study reveals clues to treating Fragile X syndrome

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Scientists have discovered how the brain can self-correct disruptions in processing, pointing the way towards possible new treatments for autism and intellectual disability.

Self-correction

Targeting faulty brain function with a new drug treatment helped the

brain to self-correct, improving cell function and reducing seizures in tests with mice.

The findings by researchers studying Fragile X syndrome – an inherited form of autism – could aid development of therapies and add to the understanding of the condition, which causes learning disabilities and seizures.

Mouse model

Scientists made their discovery using mice whose genetic make-up mirrored that seen in the DNA of people with the syndrome.

Researchers discovered that enhancing a brain receptor – known as the muscarinic acetylcholine receptor M4 – with drugs led to normalised brain activity and reduced seizures in mice.

The study, led by scientists at the University of Edinburgh's Patrick Wild Centre, focused on an area of the brain known as the hippocampus, which is linked to learning and memory.

Treatments

Experts say the findings open new avenues toward developing drug therapies and may shed light on why existing approaches to treatments have failed.

Fragile X syndrome affects about one in 4,000 boys and one in 6,000 girls in the UK. There are no treatments available to overcome the associated learning difficulties.

"Our findings give us insights into how Fragile X syndrome affects the

brain on a cellular level. Our next steps will be to understand more about the role of the M4 receptor in brain signalling in Fragile X, and its potential role in future drug development studies," says Dr Emily Osterweil.

More information: Sophie R. Thomson et al. Cell-Type-Specific Translation Profiling Reveals a Novel Strategy for Treating Fragile X Syndrome, *Neuron* (2017). DOI: 10.1016/j.neuron.2017.07.013

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