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Research unveils new hope for deadly childhood disease

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Investigators at the University of Rochester Medical Center have uncovered a promising drug therapy that offers a ray of hope for children with Batten disease – a rare neurodegenerative disease that strikes seemingly healthy kids, progressively robs them of their abilities to see, reason and move, and ultimately kills them in their young twenties.

The study, highlighted in the January edition of *Experimental Neurology*, explains how investigators improved the motor skills of feeble mice that model the disease, helping them to better their scores on successive coordination tests.

“No treatment currently exists for these kids – nothing to halt the disease, or even to slow it down,” said one of the study’s authors, David Pearce, Ph.D., a nationally renowned Batten disease expert and biochemist at the University of Rochester. His team has published more than 50 studies on the disease’s basic mechanisms.

“Since deterioration of motor skills is the rule – in fact, it’s one of the primary symptoms in children with the disease – the idea that these functions might be able to be partially restored or improved is groundbreaking,” Pearce said.

Last year, University of Rochester researchers discovered that, in mice with the disease, a set of the brain’s receptor cells – known as the AMPA receptors – were unusually sensitive to glutamate, a neurotransmitter

vital for learning and memory. These ‘super-ticklish’ receptors were located in the cerebellum, a brain region that plays a hefty role in sensory perception and motor control.

“For us, their abnormal activity made them key suspects in the brain dysfunction and neurological decline associated with the disease,” Pearce said.

To test that, researchers administered a drug that partially blocks these receptors and dims their activity.

Impressively, when diseased mice that received the drug, they – for the first time – became able to better their scores on successive coordination tests.

And, though they never reached the same level of nimbleness as healthy mice did, they were fierce candidates for the title of “most improved players.” Over the course of the testing, they achieved nearly the same degree of improvement in their before and after coordination scores as healthy mice did. In fact, almost second for second.

“It seems we may have corrected some sort of motor learning deficit in the diseased mice,” Pearce said.

While optimistic about these findings, Pearce stressed the importance of reminding affected families that this work is preliminary.

“Much research is yet needed,” Pearce said. “The prospect of offering this sort of investigational medicine to affected children is still years out.”

Still, he is further encouraged that a drug called Talampanel – very similar to the blocking compound used by his team in Rochester – is

currently in phase II clinical trials for treating epileptic seizures.

Unlike most anticonvulsants, which typically target cells known as NMDA receptors, Talampanel works by partially blocking AMPA receptors.

Source: University of Rochester