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Hope raised for reversing severe childhood disease

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By Will Dunham

WASHINGTON (Reuters) - Scientists searching for a way to treat the rare but severe childhood neurological disorder Rett syndrome have reversed the disease in mice, raising hopes for doing the same in people.

In a study appearing on Thursday in the journal *Science*, researchers led by Adrian Bird of the University of Edinburgh in Scotland switched on a gene called MECP2 in mice with the equivalent of Rett syndrome to make their symptoms vanish.

The surprising results contradicted the notion that damage to the brain caused by the disease, which occurs nearly exclusively in girls, is permanent.

"It rocked us back on our heels because in a way we were expecting a more disappointing result," Bird said in an interview.

About one in every 10,000 to 15,000 girls born have Rett syndrome, which affects all racial and ethnic groups worldwide, according to the U.S. National Institutes of Health.

In their first six to 18 months of life, the children develop apparently normally before the onset of devastating symptoms. Children with Rett syndrome commonly show autistic-like behaviors in the early stages.

It destroys speech, normal movement and functional hand use, and causes breathing difficulties, susceptibility to epileptic fits and tremors like those in Parkinson's disease. Many patients are confined to wheelchairs. Those who can walk do so with an abnormal, stiff-legged gait.

The disease, first described by an Austrian doctor in 1966, is caused by mutations in the MECP2 gene.

The researchers made the gene fully functional over a four-week span. This eliminated the mice's tremors, returned their breathing to normal and normalized their mobility and gait -- even in animals just days away from dying. Bird's team also reactivated the gene in mice before the disease was apparent and prevented the development of symptoms.

'HUNKY-DORY'

"It's a real reversal, and they (the mice) go back virtually to normal," Bird said. "There are a few vestigial problems, but they're very minor, like they walk in a slightly funny way. But by and large everything seems to be hunky-dory after we've done this."

Bird, who also heads the Rett syndrome Research Foundation's scientific advisory board, cautioned that producing a therapy to cure people is "more than a few steps away."

The MECP2 gene holds instructions for making a protein that acts as one of

many biochemical switches that prompt other genes to turn off and stop producing their own proteins. Mutations in this gene also are present in some childhood schizophrenia, autism and learning disabilities.

A key finding in the study, Bird said, is that the damage in Rett syndrome is not irrevocable.

"It is an ingrained view in most people that once there are symptoms to do with the brain that it's too late to do anything about them. But in a case where all the neurons are still alive and they've not been lost -- which is the case in Rett syndrome and autism and schizophrenia -- that's not a foregone conclusion," Bird said.

Dr. Huda Zoghbi, a noted researcher into Rett syndrome at Baylor College of Medicine in Texas, called the findings "extraordinary."

"The successful restoration of normal function demonstrated in the mouse models suggests that if we can develop therapies to address the loss of MECP2, we may be able to reverse neurological damage in children and adults with Rett, autism and related neuropsychiatric disorders," Zoghbi said.

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