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Study offers treatment hope for Rett syndrome

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The symptoms of a severe autism spectrum disorder affecting at least 10,000 children in the UK could be reversed following research by Scottish scientists.

Rett syndrome, which can leave children wheelchair bound, unable to speak and suffering from breathing difficulties, has until now been considered an untreatable neurological disorder.

But experts have now been able to make symptoms disappear in mice by activating a specific gene.

The research, led by the University of Edinburgh in collaboration with the University of Glasgow, focused on the behaviour of the MECP2 gene that causes the syndrome to develop. They found that when this gene was activated in mice, which had previously been born with it switched off, symptoms such as breathing and mobility difficulties ceased. Over a four week period, the mice often became undistinguishable from healthy counterparts.

The findings give impetus to ways of treating Rett syndrome, which mainly affects girls. Further research may also show that the results could apply to other autism spectrum disorders.

Prof Adrian Bird, Director of the Wellcome Trust Centre for Cell Biology at the University of Edinburgh, led the research and first discovered the MECP2 gene in 1990. He said: "The results we came

across were entirely unexpected. Until now it had been thought that Rett syndrome is irrevocable, but our findings show that the damage to nerve cell function is, in fact, reversible. This gives a major boost to the search for treatments or a potential cure."

The research, which is published online by the journal Science in Science Express, was funded by the Wellcome Trust, Rett Syndrome Association UK (with support from Jeans for Genes) and the US-based Rett Syndrome Research Foundation.

Although Rett Syndrome is present at birth, it becomes more evident during the second year. It is believed to be the second most common cause of severe and profound learning disability in girls.

Potential treatment, following on from the research, could range from overriding the mutated version of MECP2 with the activation of a healthy version of the gene. Alternatively, therapy could focus on drug treatment to inhibit the action of proteins expressed as a result of a MECP2 mutation.

Chris James, director of Rett Syndrome Association UK, said: "The Rett Syndrome Association UK is extremely delighted to hear about the results of Professor Bird's research. This is a very significant step on the road for future therapeutic approaches to Rett syndrome and, whilst the work in this area is still at an early stage, it will give hope to those families affected by Rett syndrome. It is particularly pleasing for us that we have been able to help fund this research that could have such a positive benefit for people with Rett syndrome, their families and carers."

Source: University of Edinburgh