Rett UK
25 years closer to the cure

Reversal of Neurological Defects in a Mouse Model of Rett Syndrome

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It was a fairly obvious experiment to do, said Adrian Bird, PhD, who led the study. Once you know it's a single-gene disorder, with late onset, the question is whether, if you turn the gene back on, can you reverse the symptoms?

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Abstract

Rett syndrome is an autism spectrum disorder caused by mosaic expression of mutant copies of the X-linked *MECP2* gene in neurons. However, neurons do not die, which suggests that this is not a neurodegenerative disorder. An important question for future therapeutic approaches to this and related disorders concerns phenotypic reversibility. Can viable but defective neurons be repaired, or is the damage done during development without normal MeCP2 irrevocable? Using a mouse model, we demonstrate robust phenotypic reversal, as activation of MeCP2 expression leads to striking loss of advanced neurological symptoms in both immature and mature adult animals.

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