

# Discovery may lead to new treatment for Rett Syndrome

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Researchers at Oregon Health & Science University have discovered that a molecule critical to the development and plasticity of nerve cells – brain-derived neurotrophic factor (BDNF) -- is severely lacking in brainstem neurons in mutations leading to Rett syndrome, a neurological developmental disorder. The finding has implications for the treatment of neurological disorders, including Rett syndrome that affects one in 10,000 baby girls.

The new discovery is published online in *Neuroscience* and is expected in the print issue of *Neuroscience* in March.

Using a mouse model of [Rett syndrome](#), the OHSU team found that mutant neurons in the brainstem fail miserably at making BDNF. When normal neurons are faced with a respiratory challenge, such as low oxygen, they dramatically increase the production of BDNF, whereas mutant neurons do not.

According to the National Institutes of Health, Rett syndrome is estimated to affect one in every 10,000 to 15,000 live births and almost exclusively girls because it is caused by an X-linked gene mutation. In addition to severe problems with motor function, other symptoms of Rett syndrome may include breathing difficulties while awake.

"The new finding, coupled with our previously published data that show BDNF is involved in normal maturation of neuronal pathways controlling cardiorespiratory function, could play a significant role in the

development of a treatment for Rett syndrome," said Agnieszka Balkowiec, M.D., Ph.D., principal investigator and associate professor of integrative biosciences in the OHSU School of Dentistry; and adjunct assistant professor of physiology and pharmacology in the OHSU School of Medicine. To conduct this research, Balkowiec partnered with John M. Bissonnette, M.D., professor of obstetrics and gynecology, and cell and developmental biology in the OHSU School of Medicine.

**More information:**

<http://www.sciencedirect.com/science/article/pii/S0306452212000395?v=s5>

Provided by Oregon Health & Science University

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