

# Researchers link early stem cell mutation to autism

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In a breakthrough scientific study published today in the *Proceedings of the National Academy of Sciences*, scientists at the Burnham Institute for Medical Research have shown that neural stem cell development may be linked to Autism. The study demonstrated that mice lacking the myocyte enhancer factor 2C (MEF2C) protein in neural stem cells had smaller brains, fewer nerve cells and showed behaviors similar to those seen in humans with a form of autism known as Rett Syndrome.

This work represents the first direct link between a developmental disorder of neural stem cells and the subsequent onset of autism.

The research team was led by Stuart A. Lipton, M.D., Ph.D., a clinical neurologist and Professor and Director of the Del E. Webb Neuroscience, Aging and Stem Cell Research Center at Burnham.

"These results give us a good hint of how to look at Rett Syndrome and potentially other forms of autism in humans," said Dr. Lipton. "Having identified a mutation that causes this defect, we can track what happens. Perhaps we can correct it in a mouse, and if so, eventually correct it in humans."

Discovered in Dr. Lipton's laboratory, MEF2C turns on specific genes which drive stem cells to become nerve cells. When MEF2C was deleted from neural stem cells in mice, there was a faulty distribution of neurons accompanied by severe developmental problems. Adult mice lacking MEF2C in their brains displayed abnormal anxiety-like behaviors,

decreased cognitive function and marked paw claspings, a behavior which may be analogous to hand wringing, a notable feature in humans with Rett syndrome.

"There's a yin and yang to this MEF2C protein," said Dr. Lipton. "My laboratory recently showed that MEF2C induces embryonic stem cells to become neurons. In this new research, we show that knocking out MEFC2 in the brain results in mice with smaller brains, fewer neurons and reduced neuronal activity. The commonality is the protein's association in making new neurons."

Rett syndrome, a form of autism, afflicts more girls than boys and results in poor brain development, repetitive hand motions, altered anxiety behaviors and the inability to speak. Patients with Rett Syndrome also suffer from seizures and other debilitating neurological symptoms.

Source: Burnham Institute

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