Medical Xpress

Stem cell research produces a key discovery for Fragile X Syndrome

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An important finding has been made by McMaster researchers about Fragile X Syndrome (FXS), a sex-linked genetic disorder that affects approximately one in 4,000 males and one in 6,000 females.

FXS is the most common genetic disorder associated with mental impairment. The affected gene (FMR1) leads to inactivation of the FMR1 gene product, known as the fragile X mental retardation protein (FMRP).

Brain development in the absence of this protein leads to cognitive effects, learning and memory problems, attention deficit, hyperactivity and autistic behaviors. Many children go undiagnosed with Fragile X.

Although the exact functions of FMRP in the brain are unresolved, there is compelling evidence that FMRP is important for normal function at the sites of communication between cells or neurons in the brain. Until now, FMRP was thought to be found only in neurons.

Stem cells are candidates for cell therapy in neurological disorders since they are capable of producing all cell types in the nervous system.

When studying the development of adult stem cells from the mouse brain, Laura Pacey, a Ph.D. student in professor Laurie Doering's laboratory, realized that cells, in addition to neurons, were also producing the FMRP. Doering is an associate professor in the Department of Pathology and Molecular Medicine.

Using specific markers to identify cell types, it was apparent that another major class of cells, called glial cells, also contained FMRP. Glial cells provide structural and metabolic support for neurons and they are necessary for normal function of the nervous system.

This discovery is important because these neuro-glial cells (astrocytes) play important roles in the development and maintenance of normal communication between neurons in the brain and spinal cord. So the absence of FMRP in astrocytes may contribute to the abnormal neuronal structures seen in the brains of Fragile X patients. The results of this research have strong implications for the cellular causes of FXS and will open new streams of research.

“This is an unexpected finding” states Doering. “Like fitting a piece of a puzzle that suddenly paints the main picture in a different perspective. We have another major cell type as a focus in Fragile X research. It will supply needed insight on the biology causing Fragile X and help to strengthen the potential for treatment strategies”.

Source: McMaster University

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