

By: Stefania 2015 Science Editor

[Sex Chromosome-Linked Brain Retard, Much More Complex Than Previously Thought](#)

The fragile X Syndrome

The sex chromosomes come with nasty effects, like the fragile X Syndrome (FXS), a genetic disease experienced by about one in 4,000 males and one in 6,000 females, and it is the most common cause of genetic mental impairment. The mutation of the FMR1 leads to the synthesis of an inactive FMR1 protein, named the fragile X mental retardation protein (FMRP). The lack of this protein impairs the brain development with severe cognitive, learning and memory issues, attention deficit, hyperactivity and autism, but many children are not detected with this severe disorder. The precise functions of FMRP in the brain are unknown, but it appears that FMRP is crucial for normal functioning at the synapses (connection sites) between brain cells (neurons) and neurons. The FMRP was found only in these neurons, till now, when a team at McMaster has discovered that mice stem cells, that can produce all cell types in the brain, produce FMRP too. Cell markers also showed that another main cell type, the glial one, also produced FMRP. Glial cells are auxiliaries of the brain, providing scaffold and food for neurons. Astrocytes, a type of glial cells, are crucial in the development and normal connection between brain neurons and spinal cord. Thus, the lack of FMRP in astrocytes can produce the impaired neuronal structures observed in the brains of Fragile X cases. "This is an unexpected finding. 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", said co-author Laurie Doering, an associate professor in the Department of Pathology and Molecular Medicine, who made the research together with Laura Pacey, a Ph.D. Student.