New Down Syndrome Protein Found

Researchers identify a protein involved in the chromosomal disorder that could explain its characteristic learning deficits.

By Edyta Zielinska | March 26, 2013

Down syndrome is caused by a triple copy of chromosome 21, which leads to a number of cognitive and physical delays. Now researchers from the Sanford-Burnham Medical Research Institute in La Jolla, California, have found a protein that restores some of the cognitive and behavioral disorders found in the disease.

Mice who were deficient in the SNX27 protein exhibited similar characteristics to mice with Down syndrome—namely, they had fewer glutamate receptors, which are important for learning and memory, the team reported in *Nature Medicine* on Sunday (March 24). The researchers also showed that in mice with Down syndrome, the protein is blocked by a molecule encoded on chromosome 21, and produced in excess in Down syndrome mice as a result of their trisomy. When the team supplemented SNX27 in the brains of mice with Down syndrome, they could see restoration of the glutamate receptors and improvements in memory deficits. “In Down’s syndrome, we believe lack of SNX27 is at least partly to blame for developmental and cognitive defects,” senior author Huaxi Xu told BBC News.

“This particular study is of interest; however, the genetic causes of Down’s syndrome are very complex and we are still a long way away from the development of therapeutic treatments that might lead to improvement to cognition in people with Down’s syndrome,” Carol Boys, chief executive of the Down’s Syndrome Association, told the BBC.

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