Researchers shed light on role of genes in autism

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(Medical Xpress)—Research carried out by Medical Research Council (MRC) researchers at the University of Oxford has uncovered a chain of genetic events that are common in individuals with autism, and have examined for the first time how this chain may influence how messages are sent between nerve cells in the brain. This knowledge will help researchers better understand the role that genetics plays in autism.

Autism Spectrum Disorders (ASD) affects around 1 per cent of the population and typically cause difficulties in social interaction, communication and repetitive behaviour. While it's known that genes can play a strong role in the development of ASD, doctors are currently only able to identify the exact genetic cause in around one in five cases.

The team, based at the MRC Functional Genomics Unit (FGU), looked at 181 autism patients who had either additional copies of some genes, or fewer copies of other genes, than people without autism. In around half of these patients, the genes whose copy count had changed were found to work together in a large biological network that plays a key role in the way in which information is passed between cells in the brain. By changing the number of copies of genes within this network, the study highlighted disturbances in those with autism in the way the information was carried across synapses in the brain.

Notably, the study also found that while some genes had gained more copies while other genes had lost copies, the final effect was predicted to be the same.

Dr Caleb Webber, lead author on the study at the MRC FGU at the University of Oxford, says: "Think of a pipe that carries water. At some points along the pipe there are genes that act as taps to let more water into the pipe. At other points genes act as holes to let some of the water out. We found that in individuals with autism the mutations in all these different types of genes act in the same way to affect waterflow. This indicates the 'tap' genes are duplicated in some individuals with autism which increases flow into the pipe, while in other individuals with autism the 'hole' genes are deleted which decreases the amount of water leaving the pipe. Both of these events cause the same thing; too much water flowing through the pipe.

"Knowing not just which 'pipes' in the cell are affected in autism but also in what way they are affected helps us to know in which way we have to change the flow to restore the balance."

Professor Hugh Perry, chair of the MRC's Neurosciences and Mental Health Board, says: "Autism Spectrum Disorders are extremely complex in the way they can influence a person's ability to communicate or interact with their environment. Tracking down and understanding the functions of genes that regulate how information is passed around the brain is a crucial part of the story and will help to underpin the evidence with which diagnoses and treatments are given. This study is a clear example of how MRC-funding can use genetic studies to improve our understanding of the brain and its networks."

The team's findings are published in the journal *PLOS Genetics*.

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