Disturbances in certain genes play a role in autism

August 16 2010

Together with colleagues from an international research group, autism researcher Christopher Gillberg of the University of Gothenburg, Sweden, has found in a new study that autism can be partially explained by abnormalities in certain genes. The group's results could, in the long run, pave the way for more appropriate treatments for autism.

Prestigious journal *Nature* is publishing an article co-authored by Christopher Gillberg of the Unit for Child and Adolescent Psychiatry at the Institute of Neuroscience and Physiology, and member of the Autism Genome Project (AGP) research group.

In the article the group reveals that a survey of 1,000 individuals with autism and 1,300 without showed that Copy Number Variants (CNVs) - sub-microscopic abnormalities in the chromosomes - are heavily over-represented in autistic people.

"Some of these are inherited, while others have appeared for the first time in the person with autism," says Gillberg. "Several of the abnormalities affect the genes that we have previously shown to be linked to autism and psychological developmental disorders".

The article stresses something that Gillberg and his colleagues have long asserted, namely that autism is partly down to a number of completely different genetic abnormalities, each of which occurs in just a small number of autistic people, but which together account for an increasing proportion of all cases, and that autism is an umbrella term for a large

number of different neurobiological conditions that have the same symptom picture.

The study also provides evidence that other genes that are important for synapse development and intra-cellular communication (communication between the nerve cells) play a role in the origin of autism in some cases. It is hoped that the link with synapse development and intra-cellular communication will pave the way for more appropriate treatment in the long term.

"Only when we know what is actually causing the autism in each case can we customise a treatment involving medication and diet," says Gillberg.

Gillberg is also one of the initiative-takers behind AGP, a group of 120 scientists from 11 different countries who have joined forces to identify the genetic factors underlying autism. He and his colleagues in the group also published, in *Nature* in 2003, 2007 and 2009, the first studies to show that the mutated genes that affect the early development of synapses also cause autism in some cases.

Provided by University of Gothenburg