DEUTSCHES KREBSFORSCHUNGSZENTRUM
IN DER HELMHOLTZ-GEMEINSCHAFT

No. 37 June 10, 2010 (Sab/Koh)

Autism: Variations in Number of Genes Characterize Genome

The genetic material of autism patients often shows a number of rare genetic alterations. Many genes have variations in the number of copies – they are multiplied or deleted. Several of the affected genes also play a role in other psychiatric developmental disorders. These results have been published by an international research consortium in the latest issue of the science journal *Nature*. In Germany, scientists of the German Cancer Research Center (Deutsches Krebsforschungszentrum, DKFZ) and Goethe University in Frankfurt/Main are participating in the project.

The “Autism Genome Project” is a consortium of 120 scientists from more than 60 research institutes in 11 countries who have been investigating the genetic causes of autism since 2003. The two participating research groups in Germany are headed by Associate Professor (PD) Dr. Sabine Klauck of the German Cancer Research Center and Professor Dr. Christine Freitag of Frankfurt University.

For their recent study, the researchers investigated the genetic material of 1,000 study subjects with an autistic disorder in search of specific genetic alterations called copy number variants. For comparison, they analyzed the genomes of 1,300 control subjects. In those affected, they more often found genomic regions to be duplicated or deleted. The consortium has published these new results in the latest issue of *Nature*. Some of those affected have inherited this alteration from their parents; however, it can also occur spontaneously.

In people with autism, gene duplication or deletion is found particularly in those genes which play a role in cell growth and neural networking. “We have found particularly many copy number variations in genes which are crucial both for the development of autism and mental impairments. This underlines the hypothesis that there are common genetic risk factors for various psychiatric developmental disorders,” said Sabine Klauck.

The results show that autism is caused by a number of rare genetic alterations, each of which affects less than one percent of the population. Christine Freitag outlines the relevance of this study: “Close investigation of the newly found genetic alterations will help us gain a better understanding of how autism develops and, possibly, find targets for therapies.”

Autism patients often inherit the disorder from their parents and typically show communication deficits, impaired social skills and stereotyped behavioral patterns. The spectrum of cognitive development ranges from above-average intelligence through to mental retardation. The condition occurs in varying degrees of severity. Therefore, the medical term “autism spectrum disorders” refers to a whole group of functional disorders with similar characteristics. It includes infantile autism, Asperger’s Syndrome and atypical autism. About one percent of the population is affected by such a disorder.

The German partners in the “Autism Genome Project” (www.autismgenome.org) have been supported by funds from the German Research Association (Deutsche Forschungsgemeinschaft) and the European Union.

A picture for this press release is available on the Internet at:
www.dkfz.de/de/presse/pressemitteilungen/2010/images/PM_Autismus.jpg
Picture source: German Cancer Research Center / Yan De Andres

D. Pinto et al : Functional impact of global rare copy number variation in autism spectrum disorders. Nature 2010, DOI: 10.1038/nature09146

The German Cancer Research Center (Deutsches Krebsforschungszentrum, DKFZ) is the largest biomedical research institute in Germany and is a member of the Helmholtz Association of National Research Centers. More than 2,000 staff members, including 850 scientists, are investigating the mechanisms of cancer and are working to identify cancer risk factors. They provide the foundations for developing novel approaches in the prevention, diagnosis, and treatment of cancer. In addition, the staff of the Cancer Information Service (KID) offers information about the widespread disease of cancer for patients, their families, and the general public. The Center is funded by the German Federal Ministry of Education and Research (90%) and the State of Baden-Württemberg (10%).

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