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Discovered. A Danish led study has as the first one found genetical differences between different types of autism. It can help the researchers with distinguishing between the diverse diagnoses and make medicine aimed at the individual.

Black box of Autism

By Kristoffer Lottrup

More than one percent of the population has Autism, and the widespread diagnosis gets great research-based attention both in Denmark and internationally. Still, we do not know much about the causes of the development disorder, which approx. 70,000 Danes are diagnosed with. Now an international research team under management by University of Aarhus and Initiative of Integrated Psychiatric Research, iPSYCH of the Lundbeck Foundation, have made an important discovery, which increases the genetical understanding of Autism. In a new extensive study, a strong team of i.e. geneticists, psychiatrists and neurologists has identified 12 genes that have significance for the development of Autism. These are so-called frequent risk genes, which are genes we all carry around. They do not in themselves have much significance, but if you have sufficiently many of them, the risk grows considerably.

>> Around 80 percent of the causes for Autism are genetic, and an exceptionally large proportion of the explanation should probably be found in these frequent risk genes. We know in advance that several rare risk genes that are completely crucial for those, who carry them, but which, on the whole, does not explain quite a lot of cases,<< Jacob Grove, Associate Professor in medical genetics at Institute for Bio medicine at University of Aarhus and first author for the new study, which on Monday was published in the scientific magazine Nature Genetics, says.

Since the first Autism Diagnosis was made in 1943, the perception of the disorder has developed significantly. As soon as in the 1960's, it came from some psycho analysts that the cause for Autism should be found in neglect of care mothers, who did not give their children enough attention – the so-called >>fridge mothers<<. Since then the research of Autism has become more biologically anchored, all while the number of Autism diagnoses have grown and grown. Today the disorder signifies a broad spectrum of so-called pervasive developmental disorders – from Apergers syndrome that is characterised of social difficulties, monotonous behaviour and limiting interest areas, for highly disabling cases of infantile autism, where the individual can be mentally impaired and completely without verbal language. The diagnosis is based on symptoms, and no medical treatment exists.

But with the finding of the frequent risk genes, the development of symptom-relieving medication has moved closer.

There are reportedly more than 1000 risk genes, which has importance for development of Autism of the individual, then the 12 newly discovered genes are just a small step on the road. Still the discovery can be described as a breakthrough, as it is the first time ever that frequent risk genes have been found.

>>This is future music, but a greater awareness of the underlying biology means that we begin to understand the processes that leads to Autism and the different types of the development disorder, << Jakob Grove says.

Specifically, the researchers have in the new study looked closer on the genome of respectively 18,381 people with autism and a control group consisting of 27,969 healthy individuals. Most of them are retrieved in Danish registers and bio banks. By scanning and

comparing the whole genome of the many examined persons, the researchers have identified five frequent risk genes, which occur substantially more often for people with Autism. Subsequently, the researchers have conducted an even wider comparison of 2119 Autists and 142,379 control individual that mainly originate from international studies. Here the researchers have also included data on related illnesses such as schizophrenia and depression. On this basis, one has found seven additional gene variants that both seem to increase the risk of autism and the other disorders. These genes should now be examined closer. All in all, the researchers have examined well over nine million frequent gene variants, and the study represent a duplication of the number of examined Autists in comparison to previous studies in the field.
>> This is the preliminary crown of the work. In the upcoming years, we can present far more frequent risk genes, and seriously draw a detailed image of the biological basis for the phenomenon, << Jakob Grove says.

>>THE ultimate perspectives in this is the development of medicine, which is based on the genetics of the individual. When we localise the frequent risk genes of Autism, we become smarter on the nuances of the different branches of Autism. It can potentially lead to the development of symptom relieving medicine focused on biological imbalances of the individual, << Anders Børglum says, who is professor of medical genetics at the Institute of Biomedicine at University of Aarhus and has been at the forefront of the new study.
Børglum, who moreover is one of the research managers in the iPSYCH-project are not afraid to call the discovery of the 12 risk genes a breakthrough and compare the development with achievements of recent years within schizophrenia.
Here one found the first frequent risk genes in 2009 and have knowledge of several hundreds today. The mapping has paved the way for attempts to develop new medicine aimed at the involved genes.
According to Anders Børglum there are good prospects for something similar in the autism area. Initially the mapping of the frequent risk genes will likely lead to more precise diagnoses within the broad Autism spectrum.
>>Currently there are several different diagnostic subgroups from severe mental impairment to well-functioning people with a high a high IQ, but with certain social difficulties. They all lie within the same broad spectrum but may be slightly different. They all lie within the same broad spectrum but can be vastly different. With the new study, we demonstrate for the first time that there is crucial difference on the genetic architecture of the diagnostic sub-groups, << Anders Børglum says.
The new study has already nuanced the picture a bit. Newer research has demonstrated a connection between genetic disposition for high intelligence and the development of Autism – a connection, which the new study also confirms. However, the researchers find that the connection does not – as previously assumed – apply to all Autist sub-groups. Thus, it is only people with Aspergers syndrome and certain kinds of infantile Autism, who on average are equipped with several of the genes concerned.
>>This discovery is literally an example on, how the net becomes denser, when we get to know the frequent risk genes, << Anders Børglum says.