

Researchers identify shared and differentiating genetic variants for ADHD and autism

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There is a large degree of overlap between the genetic causes of autism and ADHD. Now, researchers from Aarhus University have found gene variants, which increase the risk of developing only one of the diagnoses and not the other.

In the group of neurodevelopmental disorders, ADHD and autism have a number of things in common: They are two of the most common child [psychiatric diagnoses](#), both [diagnoses](#) are highly heritable, and although they differ from each other with regard to the core symptoms, autism and ADHD have a significant overlap in their underlying genetic causes.

Researchers have now identified seven genetic variants that are common to both autism and ADHD, as well as five gene variants that are specific to only one of the two diagnoses. Their study is published in *Nature Genetics*.

"We have succeeded in identifying both shared genetic risk variants and genetic variants that differentiate the two developmental disorders," says Professor Anders Børghlum of the Department of Biomedicine at Aarhus University and iPSYCH, Denmark's largest research project within psychiatry, which is behind the study.

"That means that we are beginning to understand both the [biological processes](#) that are behind the development of both diagnoses, and—as something completely new—also the processes that push the developmental disorder specifically in the direction of either autism or ADHD."

What happens in the brain?

The genetic variants affect [nerve cells](#) in the brain and the way the brain develops and communicates. It is also remarkable that some of the genetic variants identified also have an impact on people's cognitive functions in general in the population.

Specifically, the researchers can for example see that some of the genetic variants that only increase the risk of autism also increase the cognitive functions of individuals, while the complementary variants,

which only increase the risk of ADHD, generally reduce the cognitive functions of individuals.

Similarly, the researchers have identified a gene variant that increases the risk of autism and, at the same time, reduces the volume of a specific brain area in people in the general population, while the complementary [variant](#) increases the risk of ADHD and increases the volume of this brain area.

Altered diagnostic guidelines

It may seem obvious, but the study is the first in the world to show that people with both ADHD and autism are double-burdened with a genetic risk of receiving both diagnoses, whereas people who only have one of the diagnoses for the most part only bear the genetic risk variants for this one condition.

"This means, for example, that people with both diagnoses have both an equally large load of ADHD [genetic factors](#) as people who only have ADHD, and at the same time the same large load of autism genetic factors as people who only have autism. So it makes very good biological sense that some people have both diagnoses," says Anders Børglum.

The researchers analyze large datasets of genetic profiles in order to learn more about diseases and developmental disorders. This can make it possible to create more precise diagnoses and earlier interventions, and ensure that the individual patient receives the right treatment.

"The autism diagnosis is typically made before an ADHD diagnosis. So if, for example, the person is also hyperactive and finds it difficult to concentrate, this may well be slightly drowned out by the autism symptoms, and we may not see the ADHD challenges," explains Anders Børglum.

"But if we have a genetic study of a person with an autism diagnosis, and we see a major genetic load of ADHD genetics, then it may be that we should monitor that person a bit more closely. In this way, we can in the future become quicker to spot the development and give the family good tools to handle this diagnosis, too."

A few years ago—due to an official diagnosis hierarchy—it was not in principle possible to diagnose ADHD in a person who had [autism](#), he says.

"But now we have shown that people with both diagnoses are in fact double burdened with the genetic risk of both developmental disorders. There is thus a clear biological difference between whether you have both diagnoses, or just one. The study is therefore a strong biological argument for the revised diagnostic guidelines, e.g. in the American Diagnosis and Classification system for Mental Disorders (DSM-5), where it is now possible for the same person to receive both diagnoses," says Anders Børglum.

"This is the first step. Here and now, the study is relevant because it helps to create a better understanding of the causes of the two [developmental disorders](#), and in the long term, this can form the basis for better diagnostics and treatment."

More information: Manuel Mattheisen et al, Identification of shared and differentiating genetic architecture for autism spectrum disorder, attention-deficit hyperactivity disorder and case subgroups, *Nature Genetics* (2022). [DOI: 10.1038/s41588-022-01171-3](https://doi.org/10.1038/s41588-022-01171-3)

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