

Genetic analysis says no such thing as 'pure autism'

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Quinn, an autistic boy, and the line of toys he made before falling asleep. Repeatedly stacking or lining up objects is a behavior commonly associated with autism. Credit: Wikipedia.

The search for genes that contribute to the risk for autism has made tremendous strides over the past 3 years. As this field has advanced, investigators have wondered whether the diversity of clinical features across patients with autism reflects heterogeneous sources of genetic

risk.

If so, it was reasoned, then selecting a group of patients with very similar clinical features might result in a "purer", i.e., more genetically homogenous, group of patients, making it easier to find [autism](#)-related genes..

Results from a new study published in the current issue of *Biological Psychiatry* now cast the validity of this view into doubt.

A large group of collaborating scientists used data from the Simons Simplex Collection, a project that extensively characterized 2576 autism simplex families, the largest such cohort amassed to date and for which the data is now available in a permanent repository.

The availability of this vast collection allowed the researchers to create phenotypic subgroups. In addition to the whole sample, this resulted in 11 subgroups of patients with similar diagnostic, IQ and symptom profiles. They then analyzed the genotypic data in an attempt to discover common genetic variants that confer risk for [autism spectrum disorder](#).

Their results did not identify any genome-wide significant associations in the overall sample or in the phenotypic subgroups. This means that the extreme clinical variability observed among patients with autism spectrum disorder does not closely reflect common [genetic variation](#).

"This study did not provide good evidence that selecting patients with similar symptoms results in a greater ability to find autism genes," said Dr. John Krystal, Editor of *Biological Psychiatry*. "This might suggest that some of the clinical variability in autism arises from causes other than genetic vulnerability, such as epigenetic changes or other responses to the environment."

It is important to note that these results are in alignment with the recent major change to autism's diagnostic classification in the Diagnostic and Statistical Manual of Mental Disorders. In its 5th revision, several separate disorders, which included autism and Asperger's disorder, were grouped into a single category termed autism spectrum disorder. That decision was based on growing evidence that the previously-distinct disorders actually reflected a continuum of severity of the same condition.

"We hope our study is a step towards a new paradigm for studies involving the relationship between psychiatric phenotypes and genotypes. Most previous studies have sought to refine or narrow phenotypic variation irrespective of its impact on genetic variation, with the expectation that such refinement will improve detection of genetic variation increasing risk for a disorder," explained first author Dr. Pauline Chaste from Centre Hospitalier Sainte Anne in France.

"Our results motivate another route, one targeting the genetic structure of traits and, for multiple traits, their genetic correlations," Chaste added. "For autism, one important implication of our results is that our colleagues have done a very good job in first defining it; however, to make better headway on discovering genetic variation underlying risk, we believe refining phenotypes in light of their impact on genetic architecture will be essential."

More information: "A Genome-wide Association Study of Autism Using the Simons Simplex Collection: Does Reducing Phenotypic Heterogeneity in Autism Increase Genetic Homogeneity?" *Biological Psychiatry*, Volume 77, Issue 9 (May 1, 2015) [DOI: 10.1016/j.biopsych.2014.09.017](https://doi.org/10.1016/j.biopsych.2014.09.017)

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