

# Clinical histories reveal surprising evidence of multiple, distinct 'autisms'

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Credit: AI-generated image ([disclaimer](#))

(Medical Xpress)—Simple patterns can emerge from even the most chaotic, complex data.

Analyzing the [electronic medical records](#) of thousands of patients diagnosed with [autism spectrum disorder](#), or ASD, a group of

researchers at the Harvard Medical School Center for Biomedical Informatics found three distinct kinds of autism, which suggests that "autism" is perhaps an umbrella term comprising several illnesses with different genetic and environmental causes and different potential treatments.

"Rather than there being one 'autism,' these findings show that there are several autisms, each with its own specific course," said Isaac Kohane, the Lawrence J. Henderson Professor of Pediatrics at Boston Children's Hospital, co-director of the Center for Biomedical Informatics and leader of the research team. The results appeared Dec. 9 in *Pediatrics*.

By separating diagnoses out in six-month segments over the first 15 years of each child's life, the researchers found one group of patients whose autism symptoms were associated with epilepsy and other seizure disorders. In a second, ASD patients had increased rates of bowel and infectious ear and respiratory symptoms. In a third, children had higher levels of psychiatric disorders, including ADHD, depression and schizophrenia. (A fourth group of patients could not be further characterized due to statistical limitations of the available data.)

The patterns of symptoms in the first three groups also correlated with different levels of expressive language disorder, timing of developmental delays and other core diagnostic measures for ASD.

Just as an understanding of the whole suite of symptoms that accompany a fever is crucial for determining whether a patient has strep or flu, Kohane said, it's crucial to understand the entire complex of symptoms associated with the different subsets of ASD in order to advance our understanding of what causes autism.

"Doing a genetic study of 'autism' is like studying fever and looking for a single cause," Kohane said.

Since most primary caregivers might see only a few ASD patients, and since the symptoms manifest differently at various periods of the child's development, having a massive, shareable database of clinical data in searchable electronic [medical records](#) was crucial to making sense of the complex picture of autism.

In a previous study, Kohane and colleagues used the Shared Health Research Information Network (SHRINE), a web-based query tool, to analyze 15,000 [electronic health records](#) of patients with ASD from HMS-affiliated hospitals. They found that people with ASD suffer from a higher burden of seizures, psychiatric illness and gastrointestinal disorders than the general population.

While parents of children with [autism](#) and other patient advocates had long suggested that this was the case, the smaller population studies conducted without information infrastructure like SHRINE were not able to find evidence that incidences of these illnesses were significantly correlated with ASD.

This current study builds on Kohane's previous findings by showing that the related illnesses are not distributed randomly throughout the ASD population. "The next step is to look at these subgroups to search for common genetic or environmental factors," Kohane said.

Electronic medical records shared in a flexible, open-source database like SHRINE provide a bird's-eye view of the medical system that offers researchers unique insights into disease and treatment. "We spend tons of money to construct these databases and open record-sharing systems," Kohane said. "We should take advantage of all that data not just to bill people for our services, but to better understand and treat disease."

**More information:** "Comorbidity Clusters in Autism Spectrum Disorders: An Electronic Health Record Time-Series Analysis." *Finale*

Doshi-Velez, PhDa, Yaorong Ge, PhDb, and Isaac Kohane, MD, PhDa.  
*Pediatrics*, December 9, 2013. [DOI: 10.1542/peds.2013-0819](https://doi.org/10.1542/peds.2013-0819)

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