

Adults with rare genetic causes of mental health disorders more likely to have chronic illness

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Adults with specific genetic causes of mental health and other brain disorders had significantly higher rates of chronic disease and twice as

many annual emergency room visits when compared to a control group without these genetic conditions, a Geisinger study found.

Rare genetic changes, known as pathogenic copy number variants (pCNVs), have large effects on [brain function](#) and are known underlying causes of autism spectrum disorder, intellectual disability, schizophrenia, epilepsy and other [brain disorders](#). Researchers have found several dozen pCNVs associated with these conditions, but little research has been done on [healthcare utilization](#) in adults with these genetic changes.

The Geisinger research team compared a group of 928 adults who had one of these pCNVs to a control group from Geisinger's MyCode Community Health Initiative who did not have a pCNV. The study found that adults with one of these genetic conditions were more likely than the [control group](#) to have a documented neurodevelopmental or psychiatric disorder, and had higher rates of diabetes, dementia and other chronic health conditions. Adults with a pCNV also recorded twice as many annual emergency room visits.

The results were published this month in *Genetics in Medicine*.

"These findings suggest that adults with pathogenic copy number variants have poorer health and require disproportionate healthcare resources," said Brenda Finucane, M.S., professor at Geisinger's Autism & Developmental Medicine Institute and lead author of the study. "Early genetic diagnosis paired with patient-centered interventions may help to anticipate these health conditions, improve outcomes and reduce the associated economic burden for these patients."

More information: Brenda Finucane et al, Medical manifestations and health care utilization among adult MyCode participants with neurodevelopmental psychiatric copy number variants, *Genetics in Medicine* (2021). [DOI: 10.1016/j.gim.2021.11.010](https://doi.org/10.1016/j.gim.2021.11.010)

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