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

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