

# research snapshot

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## The Influence of Genes on Brain Activity

### What is this research about?

Gene mutation research helps to create knowledge of how neurodevelopmental disorders (NDD) progress within the brain. This knowledge can aid in:

- Understanding how NDD's affect a person's daily life;
- Recognizing how NDDs interact;
- Finding the best treatment for NDD symptoms.

GPHN related gene mutations are the focus of this research. Gephyrin is the protein GPHN influences. This protein controls the stability of cell structure that allows for cells to function optimally. Only cells that lower brain activity have Gephyrin present. Thus, when GPHN mutations interfere with how gephyrin functions the brain activity in these areas of the brain is impaired. GPHN mutations have been connected with NDDs in past research, but the impact of these mutations is not clear.

### What did the researchers do?

To create the sample group, 4 different patient units were explored for copy number variations (CNV) in genes that are connected to GPHN.

### What you need to know:

By learning how genes impact the brain and body, it is possible to learn how disorders are connected. This finding led researchers to question if autism spectrum disorder and seizure conditions are independent disorders.

Patient units had people who were diagnosed with ASD or schizophrenia. One patient unit had people with ASD who also had a seizure disorder. The patient unit's sizes ranged from 72 to 3704. The control group was created by pooling 3 population-based adult units. These units ranged from 2493 to 21,345. From the sample groups 6 individuals, and their families, were chosen for further exploration. These families were chosen as the person with an NDD had signs of CNV in the GPHN gene. The genetic material for both parents was on file for 5 cases, which was needed to tell if CNVs are inherited.

### What did the researchers find?

This research changed the understanding of how GPHN mutations affect brain activity. This knowledge will help to integrate GPHN into

genetic testing for NDD. It also brought insight into how the impact of not lowering brain activity is related to ASD. These findings caused the researchers to question how seizures and ASD are related. ASD and seizure disorders have been thought of as associated conditions. However, this research suggests that seizures could influence the expression of ASD.

### How can you use this research?

The need of understanding the genetic influence on NDDs will be made clear to policymakers. Practitioners will benefit from staying up to date on how NDDs are diagnosed and treated.

### About the Researchers

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### Keywords

Neurodevelopmental disorders, GPHN gene codes, Glycine, GABA

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