GeneDx PRAXIS

Case study

Advancing Epilepsy Research

GeneDx's Epilepsy Answers Partnership Program has expanded access to exome sequencing for patients with epilepsy, enabling earlier and more widespread identification of genetic etiologies. Through participation, Praxis Precision Medicines has been able to support increased access to clinically appropriate testing for pediatric epilepsy patients and engage clinicians caring for these patients.

The foundation: GeneDx's Epilepsy Answers Partnership Program

For patients with epilepsy, a genetic diagnosis can play a critical role in understanding disease biology. However, access to comprehensive genetic testing has historically been inconsistent, and many patients remain undiagnosed. The Epilepsy Answers, developed by GeneDx, was designed to expand access to high-quality whole exome sequencing for patients with suspected genetic etiologies.

As a participating sponsor, Praxis supported this initiative to help increase awareness of genetic testing and identification of genetic etiologies.

Through Epilepsy Answers:

- Pediatric patients were offered **whole exome sequencing (WES)**
- Testing included **proband, duo, and trio** analysis
- Clinicians could **order testing within existing workflows**, reducing friction

GeneDx enabled **connection to providers**, supporting education and awareness of relevant research opportunities.

Complementary outreach to support awareness

In parallel with Epilepsy Answers, outreach initiatives were conducted to raise awareness among clinicians and families about genetic testing and potential clinical trial opportunities.

These efforts complemented Epilepsy Answers by increasing awareness of genetic testing and clinical research opportunities.

Impact: Building a genetically defined patient ecosystem

1 Expanded access to genetic testing and diagnosis

Through Epilepsy Answers and associated outreach:

- 3,851 exomes were sequenced
- 41% overall molecular diagnostic yield
  (includes definitive molecular diagnoses and possible molecular diagnoses)

This work significantly expanded access to genetic testing, allowed for increased understanding of disease populations, and enabled advancement of epilepsy research.

2 Strengthened clinician engagement

A key differentiator of Epilepsy Answers is its ability to bring clinicians into the process, connecting genetic insights with the providers actively managing patient care.

Through this model, Praxis gained increased visibility and connection to clinicians treating patients, supporting more informed clinical development efforts.

Why this worked:

This partnership succeeded because of the mutual shared interest to advance pediatric epilepsy care and research.

- **GeneDx** operationalized expanded access to exome sequencing and enabled connection to clinicians
- **Praxis brought a research interest in expanding the understanding of genetic etiologies across the epilepsy landscape**

Together, this created a model centered on **access → diagnosis → clinician engagement** supporting a more informed and efficient path toward advancing epilepsy research.

Looking ahead: The future of epilepsy research

As research into epilepsy genetics deepens, programs that expand access to genetic testing that generate real-world insights will play an increasingly important role in advancing research.

Programs like Epilepsy Answers demonstrate the value of:

- Expanding access to high-quality genetic testing
- Building relationships with treating clinicians
- Enabling visibility into genetic etiologies

This work reflects a significant advancement in drug development to inform therapeutic innovations for genetic disorders.

Praxis Precision Medicines is a fully integrated, leading central nervous system (CNS) precision neuroscience biopharmaceutical company, translating insights from genetic epilepsies into the development of therapies for CNS disorders characterized by neuronal excitation-inhibition imbalance. Praxis is applying genetic insights to the discovery and development of therapies for rare and more prevalent neurological disorders through its proprietary small molecule platform, Cerebrum™, and antisense oligonucleotide (ASO) platform, Solidus™, using its understanding of shared biological targets and circuits in the brain. Praxis has established a diversified, multimodal CNS portfolio including multiple programs across movement disorders and epilepsy, with four late-stage product candidates.

© 2026 GeneDx, LLC. 205 & 207 Perry Parkway, Gaithersburg, MD 20877.
All Rights Reserved. GeneDx.com | MKT-00391-v1 Aug 2026