

New Patient Insights Reveal Opportunities for Rare Epilepsy Care

Patients and families reveal critical gaps and opportunities in genetic testing, diagnosis, and precision medicine

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*Jill Davies, Genome Medical
Chief Executive Officer*

Medical today announced the release of a new white paper, "[Genetic Epilepsies: Voice of the Patient and Family](#)."

The report draws on insights from patient and caregiver surveys alongside peer-reviewed research to highlight systemic barriers in the diagnosis and care of genetic epilepsies and the opportunities to address them through coordinated action across the healthcare ecosystem.

Genetic epilepsies, including developmental and epileptic encephalopathies (DEEs) such as Dravet syndrome, CDKL5

deficiency disorder, and STXBP1-related disorders, are rare conditions that often begin in infancy and affect neurological development, cognition, communication, and quality of life. The white paper identifies four key themes shaping the current landscape: prolonged diagnostic journeys, gaps in genetic testing access, disease burden extending far beyond seizures, and a rapidly advancing precision therapy pipeline that many families have still not accessed.

The findings reveal that many families face years of uncertainty before receiving a genetic diagnosis. Among those surveyed, a meaningful subset reported waiting more than five years for a molecular diagnosis, and a majority reported seeing multiple providers before receiving one.

At the same time, the report highlights strong patient demand for genetic testing and new therapies. Prospective patients seeking testing reported not knowing how to access it and cost of testing as top barriers. Meanwhile, most surveyed families expressed interest in learning about clinical trials, yet a majority said trials had never been discussed with them.

"Families living with genetic epilepsies are actively seeking answers, testing, and opportunities to participate in research," said Jill Davies, Chief Executive Officer of Genome Medical. "This white paper makes clear that the science is advancing rapidly, but the systems that connect patients to diagnosis, precision therapies, and clinical trials have not kept pace. By bringing forward the

voices and experiences of patients and caregivers, we hope to help biopharma, health systems, and testing partners build more effective, patient-centered pathways to care and innovation.”

The report also highlights the profound impact of genetic epilepsies on the entire family. Nearly three-quarters of caregivers reported moderate or significant effects on their own wellbeing, and two-thirds indicated they left the workforce or adjusted employment to accommodate caregiving — underscoring the need for outcome measures and support systems that reflect the full scope of disease burden.

As the pipeline of gene therapies, antisense oligonucleotides, and targeted treatments grows, the report outlines opportunities to improve access and accelerate research by:

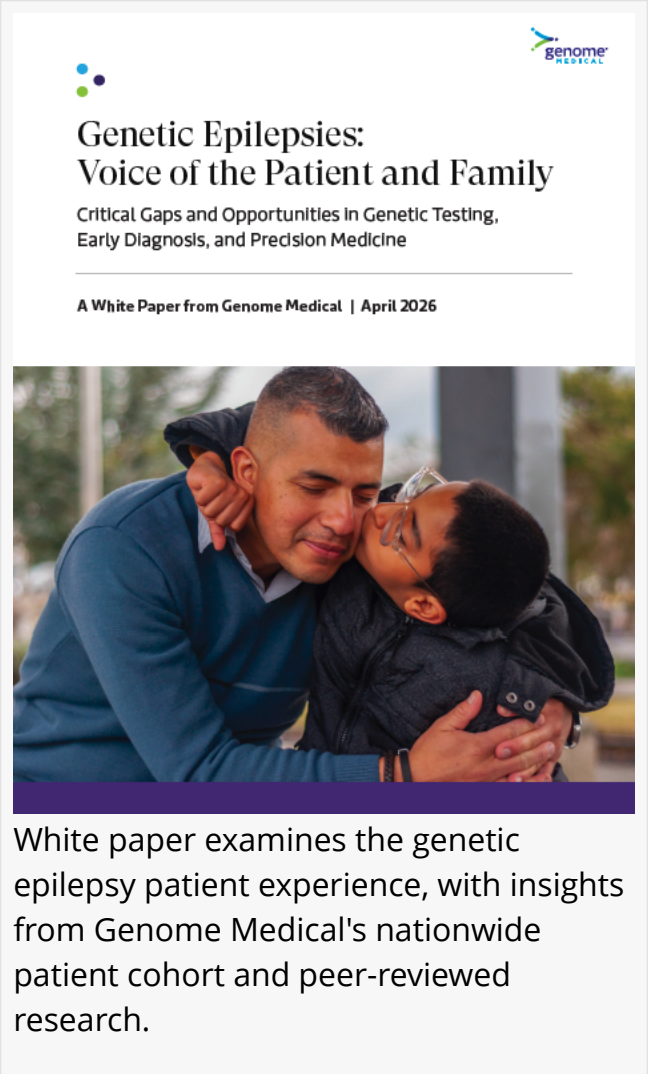
- Expanding access to genetic testing and genetic counseling
- Incorporating patient-centered endpoints that reflect developmental, cognitive, and caregiver outcomes
- Leveraging genetically characterized patient cohorts to support precision recruitment and trial design
- Implementing hybrid and family-centered clinical trial models

[Download the full white paper](#): Genetic Epilepsies: Voice of the Patient and Family

About Genome Medical

Genome Medical is transforming the landscape of rare disease care, bringing hope and answers to those navigating rare conditions. As the leading provider of telehealth-enabled genetic services, we bridge critical gaps in access, ensuring that individuals and families affected by rare conditions receive the expert guidance they need. Through timely genetic counseling, streamlined test ordering, clinical trial recruitment and patient-reported insights, we accelerate diagnoses and connect patients with potential life-changing treatments. In collaboration with life science organizations, health systems, and genetic testing labs, we are ensuring every person with a rare disease has a clearer path to care, support and breakthrough therapies. Learn more at GenomeMedical.com.

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Genetic Epilepsies: Voice of the Patient and Family

Critical Gaps and Opportunities In Genetic Testing,
Early Diagnosis, and Precision Medicine

A White Paper from Genome Medical | April 2026



White paper examines the genetic epilepsy patient experience, with insights from Genome Medical's nationwide patient cohort and peer-reviewed research.

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