



## Thank you!

The EPGP Study Team wants to extend its enormous **gratitude to you**—the international family of health care providers and research participants—who contributed your time and story to the Epilepsy Phenome/Genome Project (EPGP).

We are thrilled to report that over the **7 years** that EPGP was enrolling and phenotyping, **our final dataset contains 4,199 people with epilepsy and their family members** and more than **38,000 study activities** (interviews, EEGs, blood samples, etc) comprise the final study database—more than 6.9 million data points. Together we have worked to create, in EPGP, a scientific resource bigger and more rich than any in the world, one that is changing the way we think about genetics of epilepsy.



This effort has been a great example of how patients and health care providers can join together: 150 staff worked on EPGP at 27 clinical centers. 250 health care providers in our National Referral Network, and from around the world, referred more than 1900 families to EPGP. And more than 5,500 participants consented to be part of the EPGP research effort.

**Thank you!**

## Results, Findings, and Publications

Because of the close partnership with the international [Epi4K](#) consortium, and the efforts of the EPGP investigators, **results have been coming in!** You will be excited to hear that:

In the infantile spasms cohort, two entirely new genes have been discovered, along with many other mutations that are likely to be causing infantile spasms based on their association with epilepsy, developmental delay, other problems. These discoveries are already leading to functional analysis work—scientists are creating models of brains with these mutations, so that **new therapies can be developed**. We are really within arms' reach—maybe a few years—of **improved treatment** for infantile spasms, thanks to the collaboration and contributions of the group. In the **PMG** cohort, we have very early (unpublished) findings of at least one entirely new gene.

**Publications** have been illuminating many sides of epilepsy—from migraine to Lennox-Gastaut phenotypes. Already published papers, talks, and presentations are listed on the EPGP website [here](#). **Highlights include** (click the link to see the paper):

- [De novo mutation in epileptic Encephalopathies](#)
- [Lennox-Gastaut Syndrome of unknown cause](#)



- [Polymicrogyria-associated epilepsy](#)
- [Evidence for a shared genetic susceptibility to migraine and epilepsy](#)
- [Racial and ethnic differences](#) in epilepsy classification among probands
- [The Epilepsy Phenome/Genome Project](#)

Additional papers being written include familial patterns of epilepsy, gender differences, characteristics in status epilepticus, auras in generalized epilepsy, lateralization of epileptogenic zone, nocturnal predominance, and predictors of outcome for infantile spasms.

## Continuing of our Research Network—Together We Can!

Participants' and referring providers' continuation in future studies will be critical to the developing understanding of their epilepsy, and time is of the essence.

Through follow up “sister studies,” we are gearing up for **20 participants to have whole-genome sequencing**, with the hope that many more in the EPGP cohort will be eligible. And, as we come to understand the genes behind epilepsy and other disorders, it will be important to learn more about the other **biomarkers** (proteins and chemicals in our blood or other parts of our body) that mark the current or future progress of a disease—and to ask questions about how the epilepsy is getting better or resisting treatment for a specific family—or set of families.

By continuing to work together, we can continue to build upon the already remarkable resource that we worked so hard to build—and continue to bring light to the questions that we all ask:

- **Why do I have epilepsy?**
- **Why are my treatments working—or not making me better?**
- **Will I get better?**

New research studies continue to be developed. Please **stay in contact** with your clinical center, and look for opportunities to stay involved—through the central EPGP database or new research projects.

**Together we can help improve the lives of people living with epilepsy!**

## Contact Us

If you have any questions or suggestions, please email [Catharine.Freyer@ucsf.edu](mailto:Catharine.Freyer@ucsf.edu)

