**Why Exome Sequencing for Epilepsy is Important**

**Epilepsy is a major public health problem**

- Affects 65 million people worldwide
- 1 in 26 Americans

**Many types of epilepsy have a genetic cause**

- Gene discovery in epilepsy: Over 100 epilepsy related genes identified
- Gene found in 25-46% of unknown cases

**Exome testing to find the cause has many benefits**

- Personal usefulness to patients & families:
  - Enables risk identification in family members
  - Allows for reproductive planning
  - Ends the quest for a diagnosis
  - Ameliorates parental guilt or shame
  - Allows for connection to resources and community

- Medical usefulness to doctors:
  - In some cases, enables changes in medical management
  - Allows for prediction of epilepsy progression
  - Enables genetic counseling (many mutations shown to be de novo*)
  - Enables enrollment in clinical trials and research
  - Can decrease the time/cost of diagnostic and treatment odyssey

*De novo — A spontaneous gene alteration that arises in the developing child; not inherited.

**Exome testing can help advance research**

- Advancing our understanding of the genetic causes of epilepsy will allow us to improve the ways we anticipate, prevent, diagnose, and treat epilepsy.

- Precision therapies are already available for some patients:
  - **Gene**
  - **Treatment**
  - SLC2A1
  - Keto diet
  - POLG
  - Avoid certain medicines
  - ALDH7A1
  - Vitamin B6
  - SCN1A
  - Avoid certain medicines

**The goal:**

- Tailored individual treatments
- Enabling precision medicine in epilepsy

**About the Epilepsy Genetics Initiative (EGI)**

EGI is an initiative created to bridge the gap between people with epilepsy, clinicians, and researchers to advance precision medicine in epilepsy.

EGI has created a centralized database to hold the genetic data of people with epilepsy. The data (called exome data) will be analyzed and reanalyzed in an effort to find the cause of the person’s epilepsy (not all epilepsy has a genetic cause). Findings will be reported back to the person’s doctor. The data will also be made available to advance research.

---

**References**


---

The information contained herein is provided for general information only and does not offer medical advice or recommendations. Individuals should not rely on this information as a substitute for consultations with qualified health care professionals who are familiar with individual medical conditions and needs.