Why Exome Sequencing for Epilepsy is Important

EPILEPSY IS A MAJOR PUBLIC HEALTH PROBLEM

1 IN 26 AMERICANS

1

Many types of epilepsy have a genetic cause

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

<table>
<thead>
<tr>
<th>Gene</th>
<th>Treatment</th>
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<tbody>
<tr>
<td>SLC2A1</td>
<td>Keto diet</td>
</tr>
<tr>
<td>POLG</td>
<td>Avoid certain medicines</td>
</tr>
<tr>
<td>ALDH7A1</td>
<td>Vitamin B6</td>
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<tr>
<td>SCN1A</td>
<td>Avoid certain medicines</td>
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The goal:

Pioneering precision medicine in epilepsy

TAEIRED INDIVIDUAL TREATMENTS

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.

www.CUREepilepsy.org/EGI

The information contained herein is provided for general information only and does not constitute medical advice. Individuals should consult with qualified healthcare professionals who are familiar with individual medical conditions and needs.