

## Background

- Developmental and epileptic encephalopathies (DEEs) are characterized by early-onset seizures and developmental delays.
- Genetic testing increasingly guides diagnosis, management, and eligibility for emerging targeted therapies and clinical trials.
- Most published DEE cohorts are derived from specialty centers, which may be impacted by ascertainment bias.

### Aim

To describe a nationwide cohort of patients with disease-causing genotypes in genes associated with DEEs.

## Methods



Retrospective chart review



Patients referred to a nationwide telehealth genetic counseling practice from 2019 to 2025

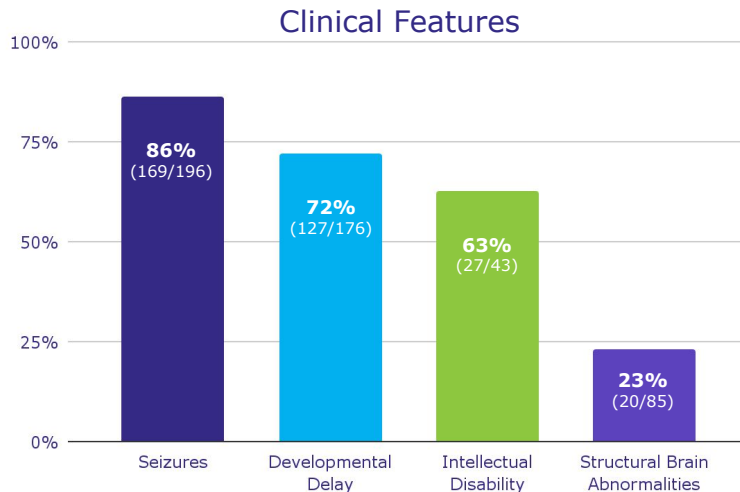


Included patients with a disease-causing genotype in one of 179 DEE genes



Chart review to extract DEE phenotype data

# Nationwide real-world cohort highlights clinical & genetic heterogeneity of developmental and epileptic encephalopathies (DEEs)



## Characteristics of a Nationwide Cohort of Patients with Developmental and Epileptic Encephalopathy Disease-Causing Genotypes

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## Results

n	Sex	Age Group
<b>269</b> patients	<b>54.6%</b> female (147/269)	<b>70.6%</b> pediatric (190/269)

Median age for children: **3y**  
(IQR 1-7y)

Patients from **46** US states.

Most frequently:



- Texas (10%)
- California (9%)
- Florida (7%)
- Indiana (6%)
- Georgia (6%)

**55.6%** (65/117) of patients with seizures had their first seizure at **<1 year old**

**61.5%** had a **de novo** variant (40/65 with parental testing)

Disease-causing genotypes in **66/179 genes**. Most frequently:



- *SCN1A* (10.0%)
- *CACNA1A* (5.9%)
- *TSC2* (5.6%)
- *MECP2* (4.1%)

Median time from first seizure to molecular diagnosis was **1y**  
(IQR 0-6y)

## Conclusion

- Characterization of DEE cohorts is increasingly relevant as genetic diagnoses inform precision care

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