A Rare Presentation of Adult-Onset KCNQ2 Encephalopathy in a Young Male with Recurrent Seizures and Encephalopathy

Dr Harsha Burgula, Dr Mridula Singh, Dr Siddharth Maheshwari, Dr Rashmi Mishra, Dr Rajinder K Dhamija
Department of Neurology, Institute of Human Behavior and Allied Sciences, New Delhi

AIMS & BACKGROUND

KCNQ2 gene mutations → Well-recognized causes of Neonatal-onset epileptic encephalopathy
Benign familial neonatal seizures (BFNS)[1]

Typical presentation: Early infancy

Case uniqueness:25-year-old male
Recurrent encephalopathy and seizures

Diagnosis: KCNQ2 encephalopathy

Aim of report:
Highlight **under-recognized adult-onset phenotype**
Emphasize role of **genetic testing** in atypical epilepsy

METHODOLOGY

A Case study of KCNQ2 Encephalopathy.

CASE REPORT

25-year-old male, software engineer

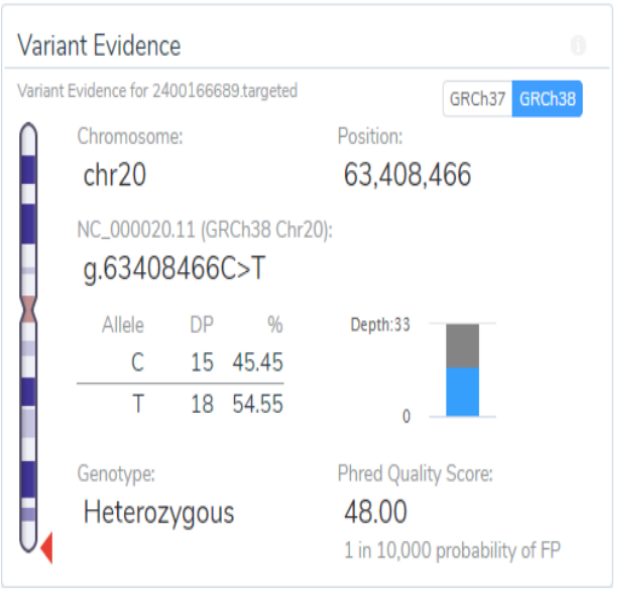
Background:
Normal birth history
No developmental delays
No family history of epilepsy

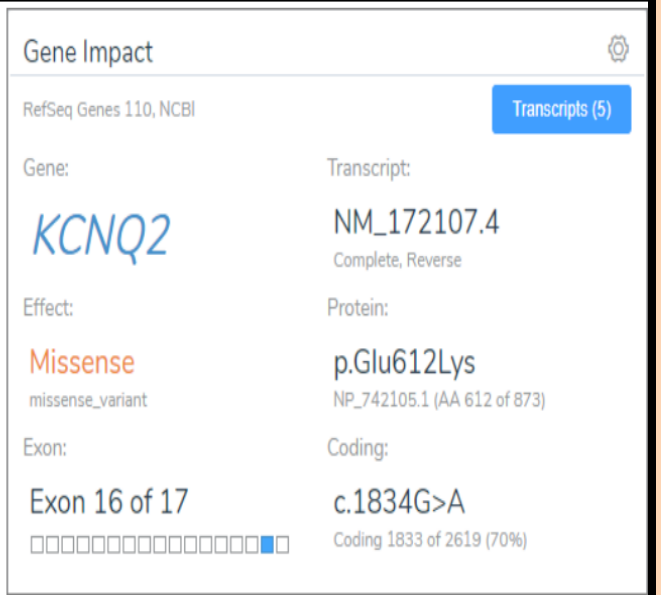
Clinical features:
Multiple stereotyped episodes of:
Abnormal behavior
Seizures

Encephalopathy requiring intubation
Each episode resolved in **7–10 days** with supportive care + anti-seizure meds

Seizure semiology:
Generalized tonic-clonic seizures (GTCS)
Focal motor seizures with impaired consciousness
Course: Very high seizure frequency

Drug-resistant: not controlled despite multiple anti-seizure medications at maximum tolerated doses





INVESTIGATIONS

Serial EEGs showed generalized slowing, Neuroimaging was normal. Genetic testing revealed KCNQ2 gene missense mutation, establishing the diagnosis.

DISCUSSION

- **Case significance**: Atypical, adult-onset presentation of **KCNQ2 encephalopathy**
- **Extension of spectrum**: Beyond usual childhood-onset forms
- **Typical adult phenotypes**: Motor, behavioral, and language issues
- **Current case features**:
- Adult-onset drug-resistant epilepsy
- Recurrent encephalopathy episodes **before seizures**
- **Normal IQ**, no developmental delay, no motor abnormalities
- **Clinical relevance**:
- Recognizing such patterns → timely genetic evaluation
- Avoids unnecessary interventions
- **Therapeutic implications**:
  - **Injectable pyridoxine**
  - **Sodium channel blockers** effective in KCNQ2 encephalopathy

CONCLUSION

KCNQ2 encephalopathy should be considered in adults with recurrent unexplained seizures and encephalopathy. Early identification through genetic testing facilitates appropriate diagnosis, counseling, and potential for precision treatment.

Reference: 1.Boets S, Hully M, Lesca G, Lagae L. Adult phenotype of KCNQ2 encephalopathy. J Med Genet. 2022;59(6):528-35. doi:10.1136/jmedgenet-2020-107449.