

# A novel mutation in *KCNQ3*-related benign familial neonatal epilepsy: electroclinical features and neurodevelopmental outcome

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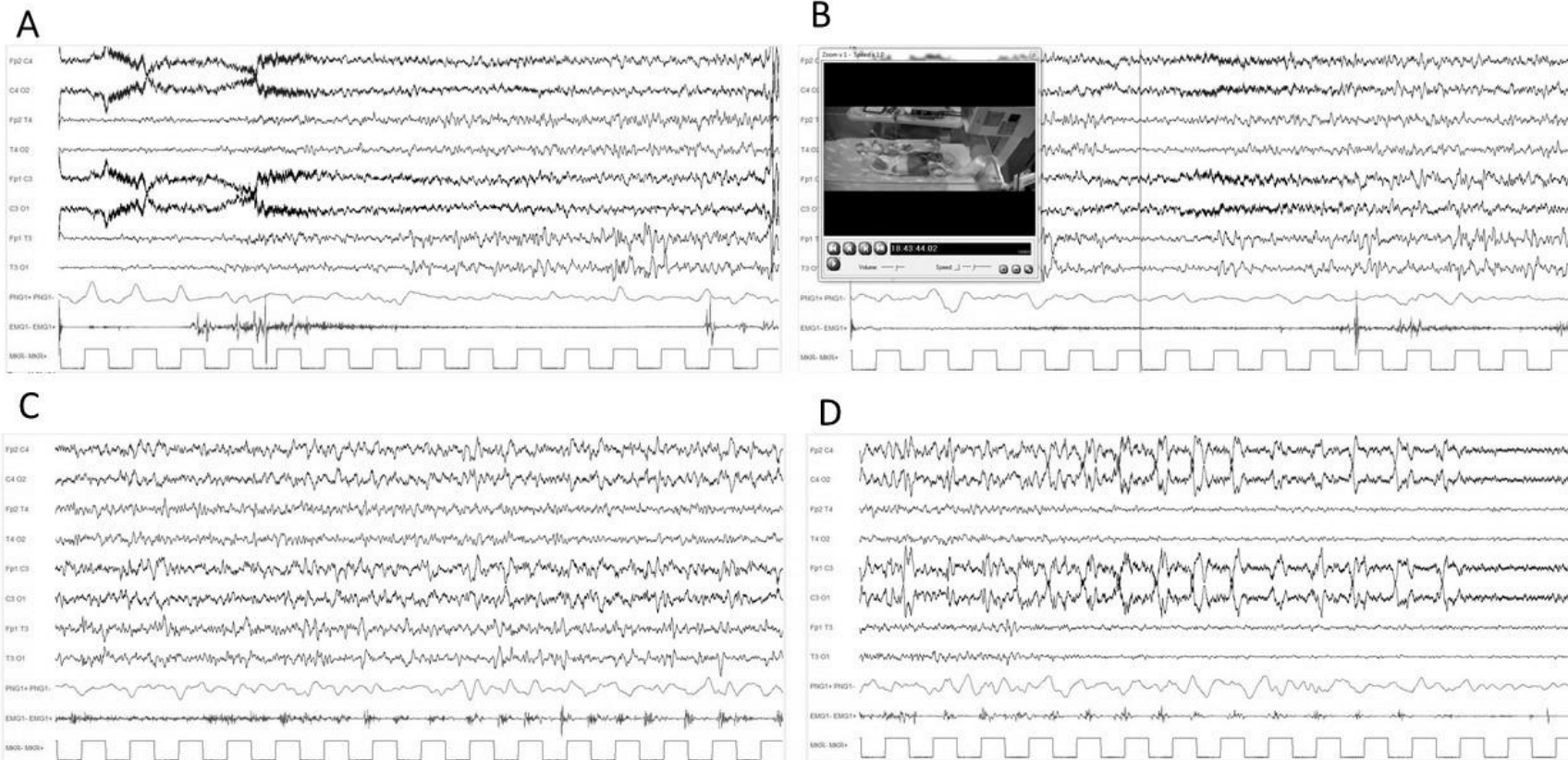
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# Benign Familial Neonatal Epilepsy (BFNE)

- BFNE is characterized by frequent clusters of unprovoked seizures starting around the second/third day of life.
- BFNE in our patient is inherited as an autosomal dominant epilepsy syndrome with incomplete penetrance and caused by a novel *KCNQ3* (c.914A>T; p.Asp305Val) variant with a favourable neurodevelopmental outcome.
- Video-EEG is the better diagnostic method to identify the electroclinical pattern.
- Video-EEG shows that the seizures are usually characterized by asymmetric tonic posturing, associated with a generalized decrease in EEG amplitude, and followed by bilateral asynchronous clonic movements and electroencephalographic bicentral sharp-wave discharges.

Electroclinical phenotype of the presented patient with BFNE. The seizure, following active sleep and arousal (**A**), is characterized by tonic extension and adduction of the upper limbs, flexion of the lower limbs, left trunk and head rotation associated with shallow breathing, tachycardia, and generalized decrease in EEG amplitude (**B**). The tonic seizure evolves into a vibratory phase (**C**), and then gradually into bilateral asynchronous clonic movements of the limbs and ocular-facial regions associated with bicentral sharp-wave discharges (**D**).



# Benign Familial Neonatal Epilepsy (BFNE)

- Our patient showed a rapid and safe response to low-dose of Levetiracetam with total seizure control. At 8 months of age the baby showed a global normal development with regular social contact without seizure relapse, and thus oral Levetiracetam was gradually withdrawn.
- We think that Levetiracetam may be included among the first-line drugs for BFNE treatment as recently recommended for Carbamazepine and other sodium channel blockers.
- The clinical course of our patient suggests that identifying the electroclinic pattern of BFNE aims to decrease the costs of hospitalization and may protect the newborn and his family from stressful situations.