CACNA1A variants have been associated with a wide range of neurodevelopmental and neurological difficulties. We all have two copies of the CACNA1A gene. Most people with a CACNA1A-related disorder have one CACNA1A variant and one normal copy of the gene. Much more rarely, variants are present in both copies, and this is called biallelic. We wanted to understand more about the range of characteristics present in individuals with biallelic CACNA1A variants to address the challenges highlighted in genetic counselling and clinical management. To do this, we summarised previous research literature and reported a new case of a child with biallelic CACNA1A variants.

What did we find?

- Biallelic CACNA1A variants are associated with different forms of epilepsies, including developmental and epileptic encephalopathy, status epilepticus and tonic-clonic seizures.
- Most individuals display motor, visual and communication difficulties as well as changes in brain structure.
- There is also variability in the characteristics present in parents carrying one of the two variants, with parents displaying none to severe neurodevelopmental difficulties.

How can this research help?

- The variability in individuals with biallelic CACNA1A variants, and family members who may carry one of the two variants, complicates genetic counselling.
- Recognition of the range of difficulties experienced by these patients and families can improve care after diagnosis.

What is next?

- Research is trying to bridge the gap between CACNA1A variants and the variability in neurodevelopmental and neurological characteristics present.
- We hope that this will improve symptom management and long-term outcomes for these rare children and families.

Resources

- You can read the full paper [here](#).
- CACNA1A-specific resources:
  - [CACNA1A-related disorders guide](#)
  - [CACNA1A foundation](#)