Clinical commentary

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# A Chinese patient with epilepsy and *WWOX* compound heterozygous mutations

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# *WWOX* gene

- The WW domain-containing oxidoreductase (*WWOX*) gene was first located at 16q21-q23 in a large family with four affected children in Saudi Arabia.
- This gene contains 9 exons, encoding 414 amino acids.
- The protein is widely present and highly expressed in the prostate, bones, lungs, endocrine tissues, and brain.
- Early infantile epileptic encephalopathy type 28, caused by *WWOX* mutation, is a type of refractory epilepsy and a serious autosomal recessive inherited neurological disease.

# Patient data

- a male child
- Convulsions involved raising the right or left limb during sleep at 1 month of age→clusters of spasms with several spasms in each cluster at 2.5 months of age→the spasms disappeared at 9 months of age
- The patient had no marked response to antiepileptic drugs

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# Patient data

- The patient was born at 37 weeks and two days.
- The weight of the patient was 3.6 kg at birth.
- Head circumference at the age of 3 and 10 months was 38 cm and 41 cm, respectively.
- The patient was unable to follow objects but could raise his head at the age of 2 months. He was consistently unable to roll over, sit alone, or speak during his short life.
- He died of suffocation due to pneumonia at one year and 23 days of age.

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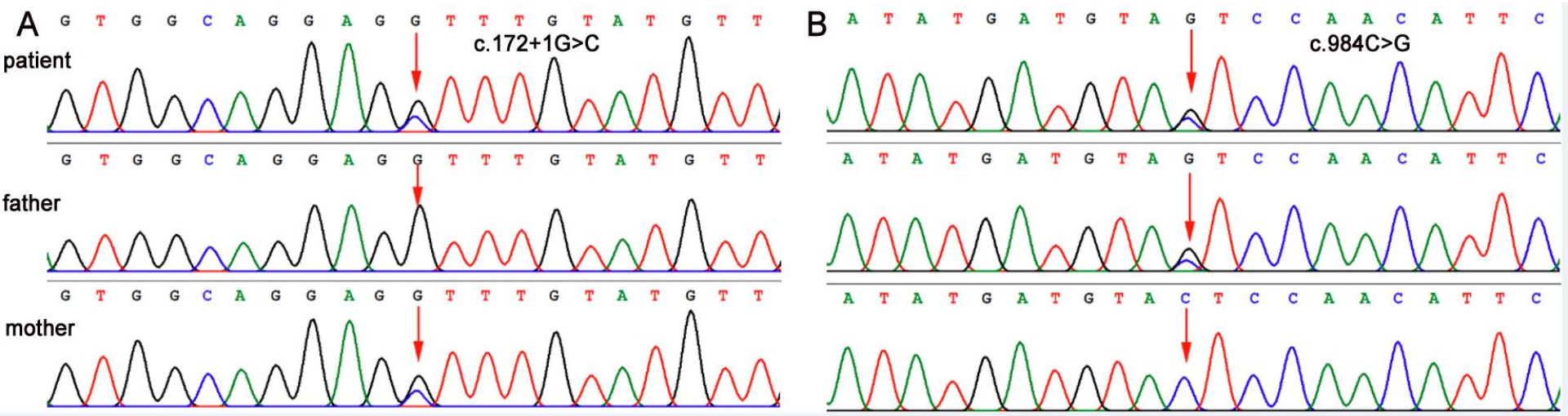
# *WWOX* gene results

*WWOX* gene sequence showing the compound heterozygous mutations (red arrows).




**A.**c.172+1G>C mutation in the patient and his mother

**B.**c.984C>G mutation in the patient and his father

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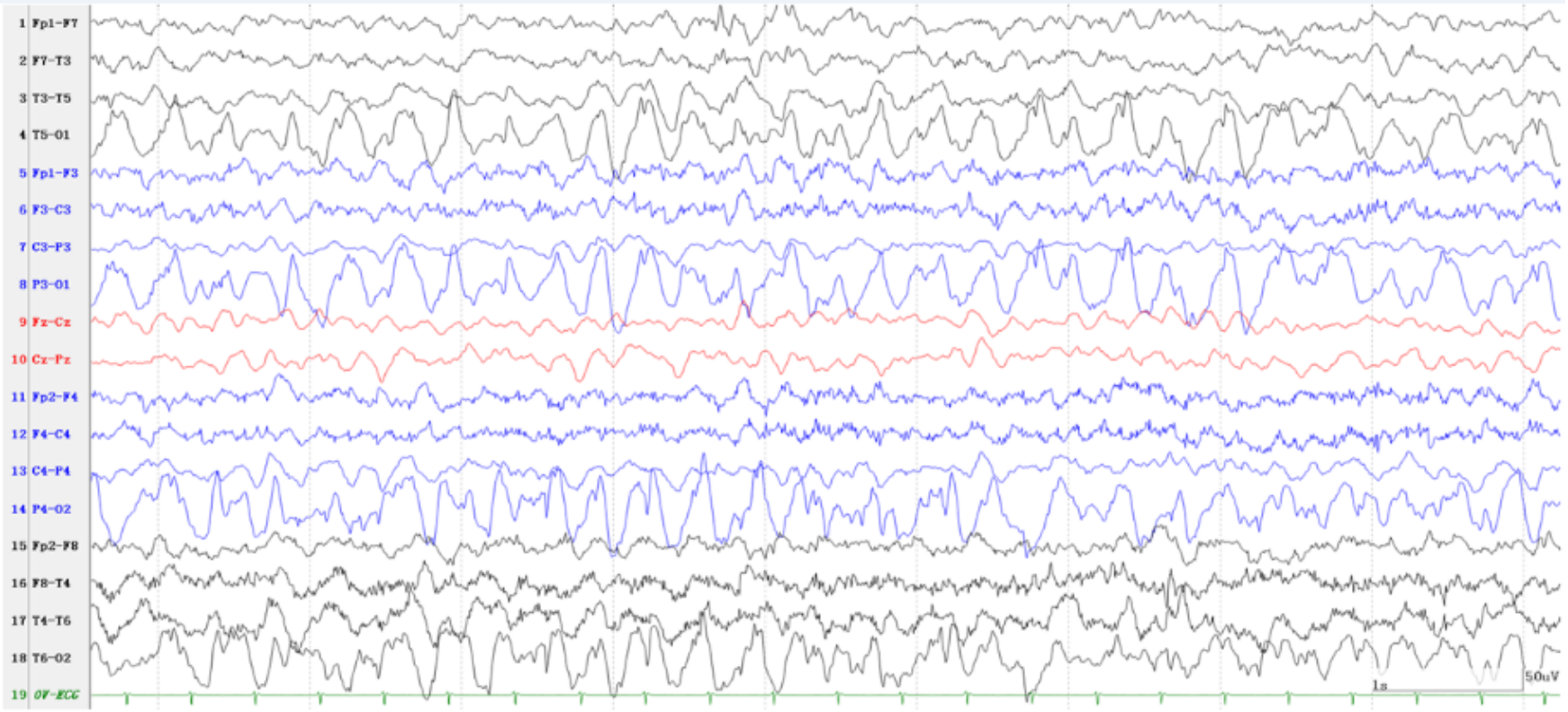
# EEG results




Background showing 4/5-Hz theta activity at the bilateral occipital region at the age of 10 months（high frequency filter = 70 Hz, low frequency filter = 1.6 Hz, sensitivity =10 uV/mm, and display speed = 30 mm/sec）

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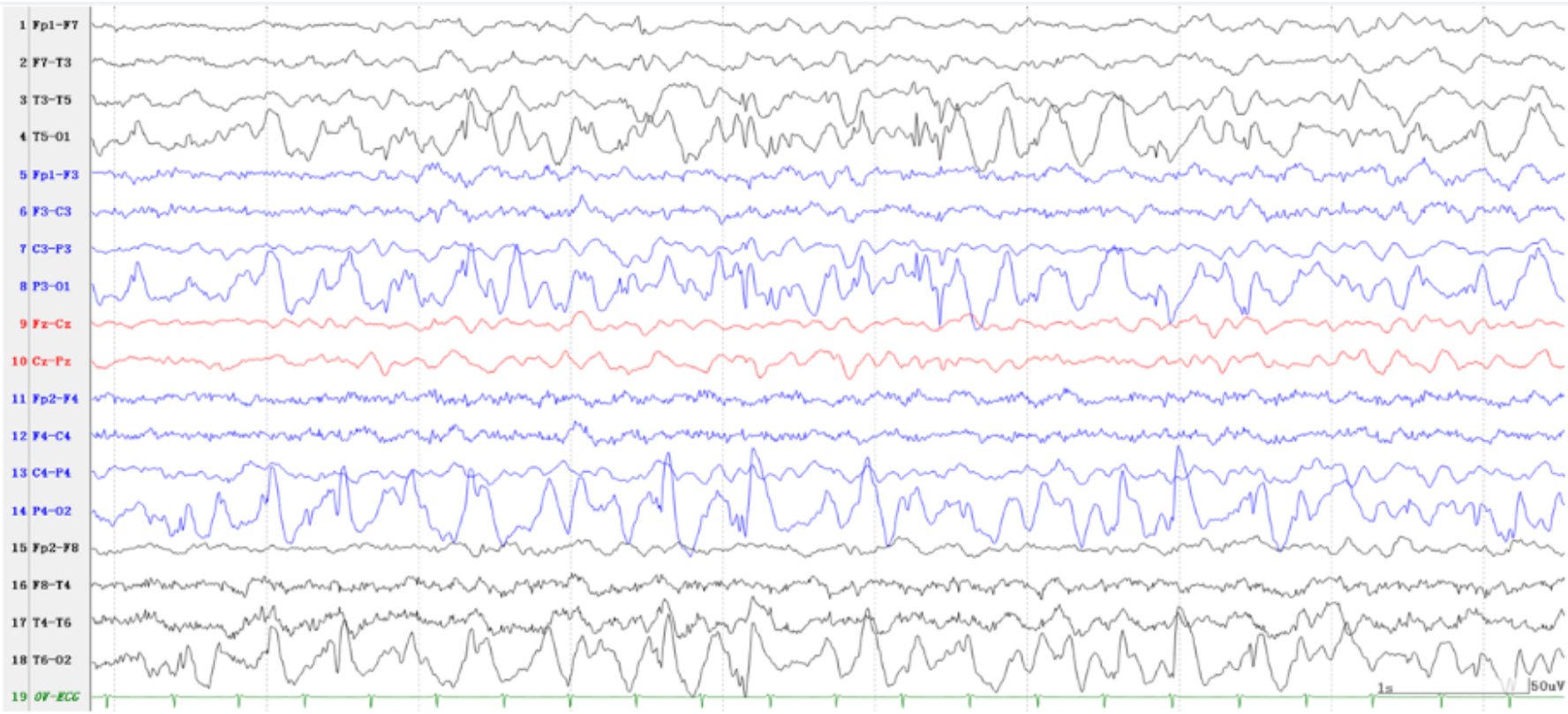
# EEG results




Interictal EEG showing spike-slow waves and polyspike-slow waves in the posterior region in both sides.

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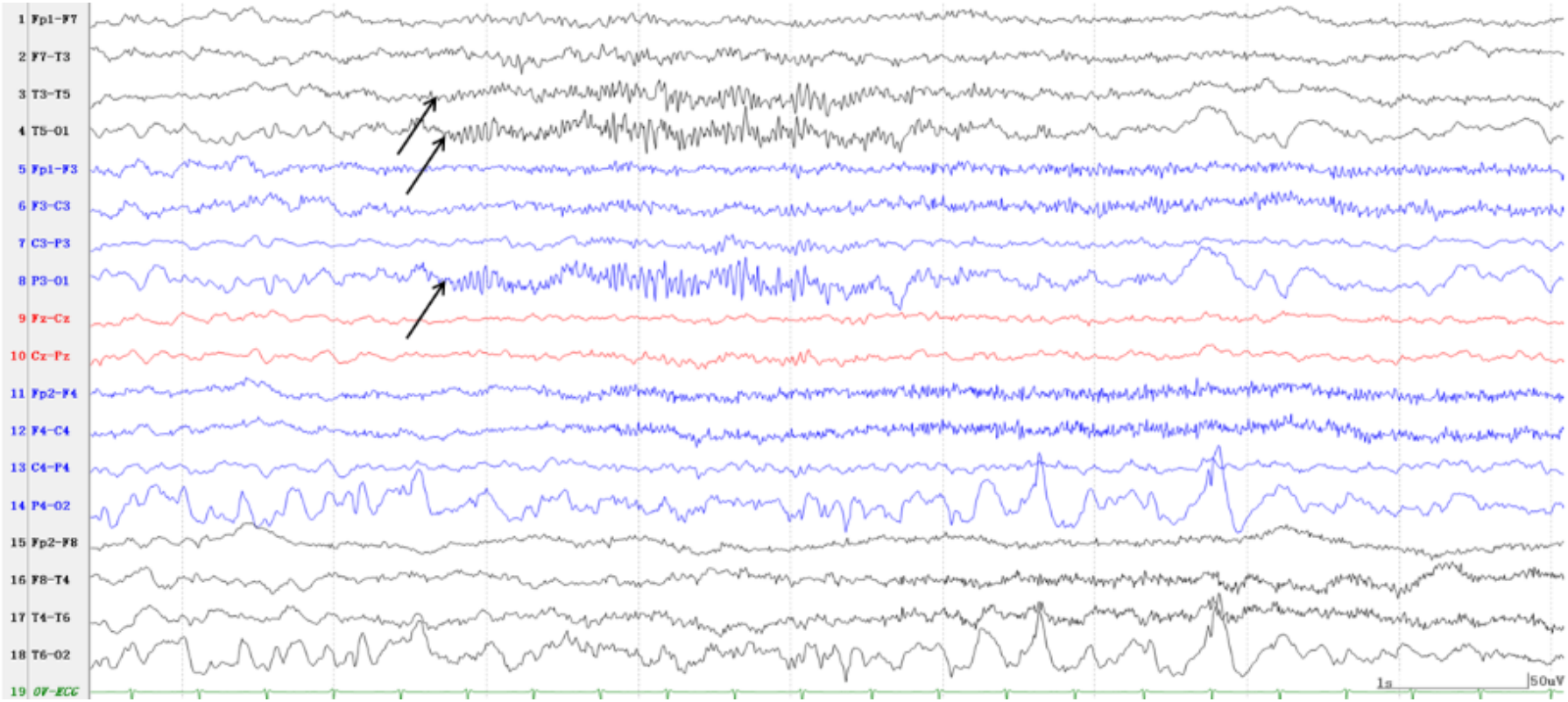
# EEG results




Ictal EEG shows a low-amplitude fast wave in the posterior region on the left side.

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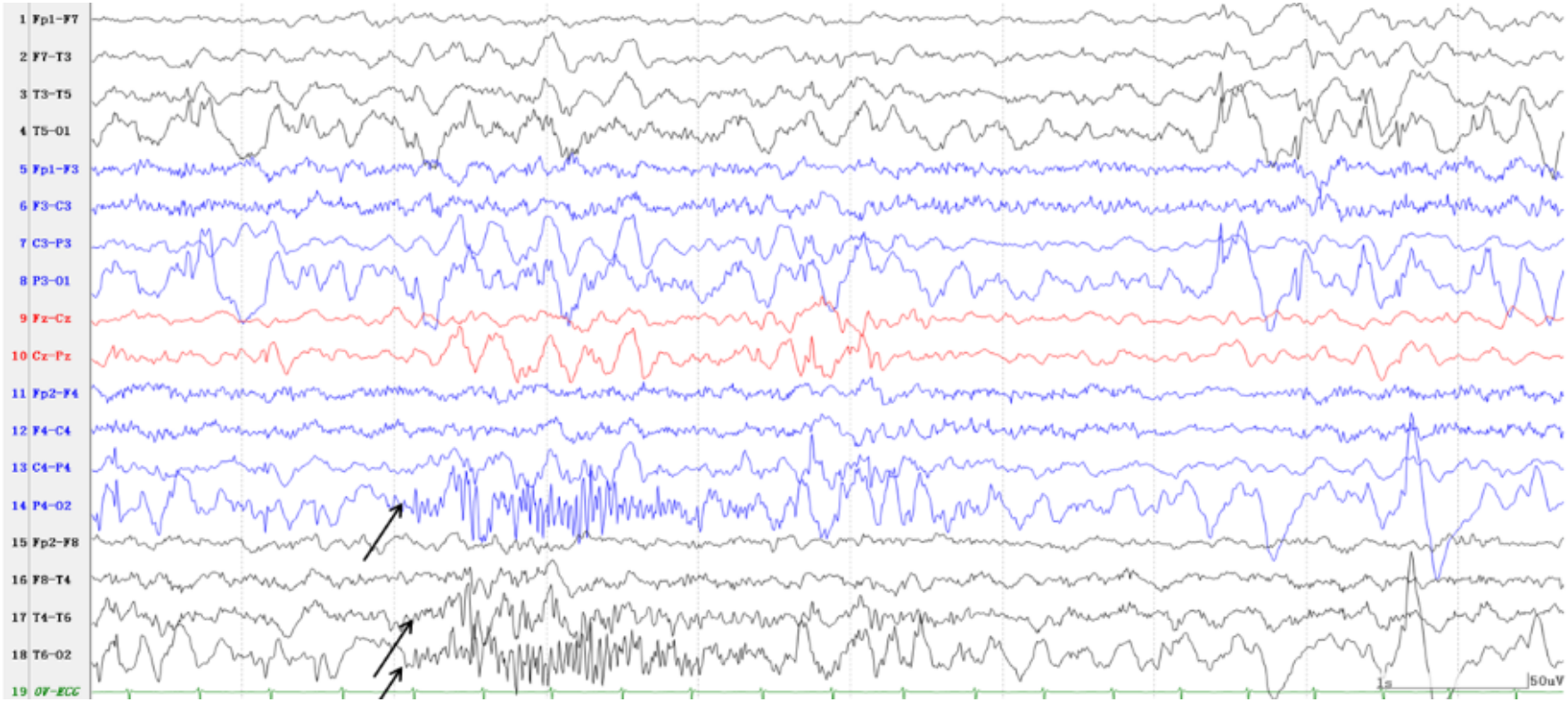
# EEG results




Ictal EEG shows a low-amplitude fast wave in the posterior region on the right side.

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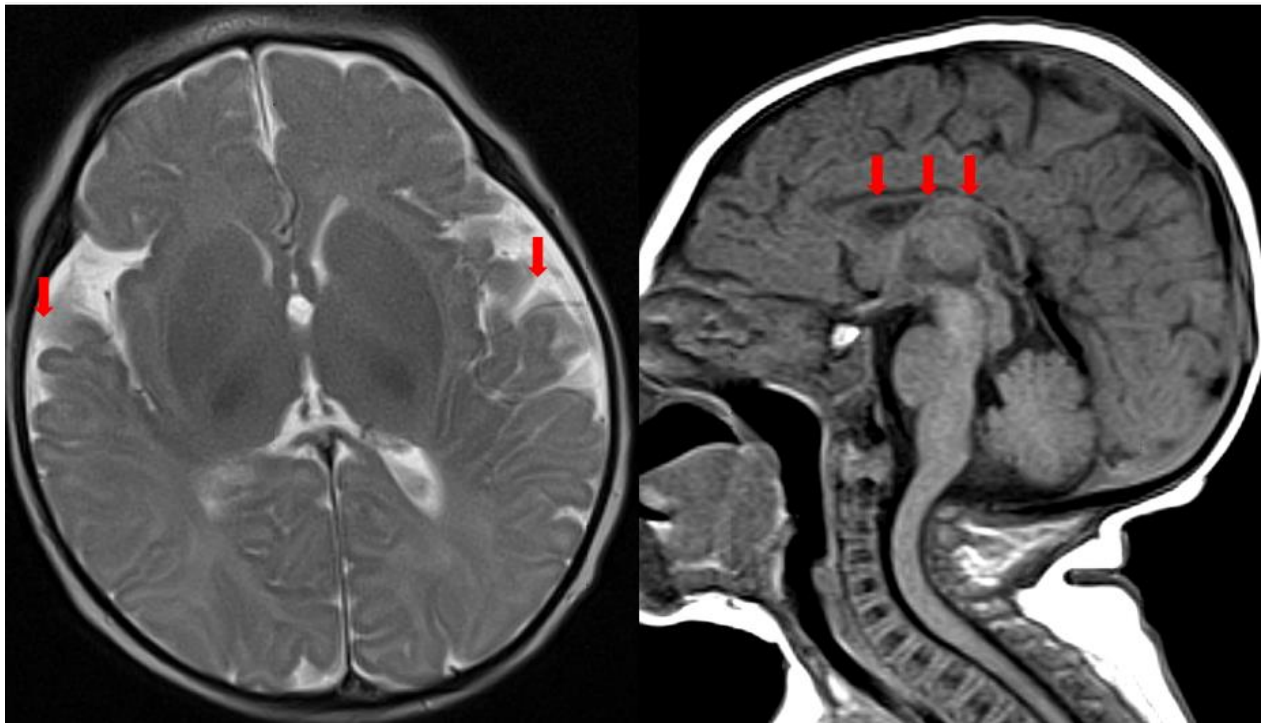

Axial T2 brain MRI showing thinning of the corpus callosum MRI.

Sagittal T1 brain MRI showing a widened subarachnoid space.

# Conclusion

- This case report adds to the list of *WWOX* gene mutations related to epilepsy.
- Future studies of patients with *WWOX* gene mutations and epilepsy should be conducted in order to further investigate genotype/phenotype correlations, and better establish the prevalence of *WWOX* gene mutations.

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