HYPOCALCEMIC SEIZURE IN A GIRL WITH FOXG1-GENE–RELATED ENCEPHALOPATHY

Magdalena Bliznakova[1], Georgi Boshev[2], Miglena Georgieva[3], Zhivka Chuperkova[3]
1 - Department of General Medicine, Medical University - Varna
2 - Graduate in Medicine, Medical University - Varna
3 - Department of Pediatrics, Medical University – Varna, Bulgaria

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Aim

The aim of this study is to present a case report of **hypocalcemia**, epilepsy and **severe vitamin D deficiency** in an 11-year-old girl with "atypical", "congenital" **Rett syndrome** due to FOXG1 gene-mutation.

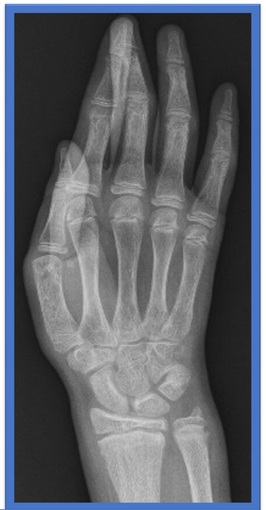
Clinical Presentation

The child had microcephaly, developmental delay, dyssomnia, and was previously diagnosed with drug resistant epilepsy with focal seizures with impaired consciousness, tonic, myoclonic and bilateral tonic-clonic seizures. She presented with multiple episodes of seizures affecting the extremities for the last 2 months, initially interpreted as epileptic seizures, and was admitted to the Children's intensive care unit of UMHAT "St. Marina" Hospital.

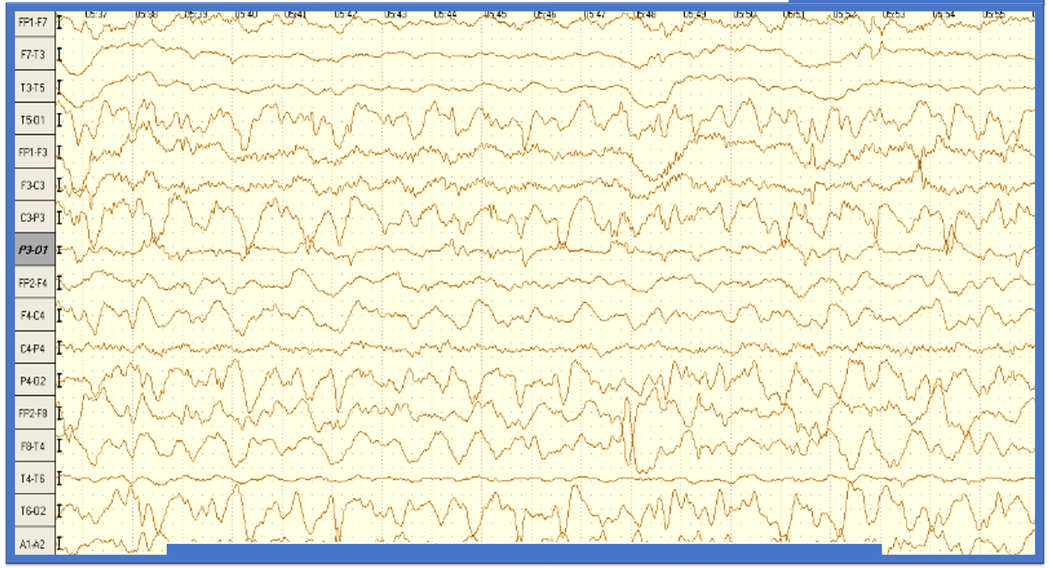
Clinical Tests

- **Hypocalcemia,** 1.3 mmol/L (ref 2.18-2.60), with ionized calcium of 0.75 mmol/L (ref 1.13-1.32).
- PTH was 511 pg/ml (ref 11-87);
- Alkaline phosphatase was 1125 U/L (ref 42-406);
- 1,25-dihydroxyvitamin D level was 5.5 pg/ml (ref. 7.61-55.5).

Hand X-Ray revealed osteoporosis.



EEG had data for diffusely delayed basal activity.



Results

Therapy with **intravenous calcium gluconate and oral vitamin D** was initiated immediately. The condition as well as the patient's general condition improved significantly and good seizure control was achieved.

Conclusion

Vitamin D deficiency which may lead to severe hypocalcaemia **is prevalent in girls with "atypical" Rett syndrome.** The presented clinical case emphasizes the importance of **hypocalcaemia as a triggering factor for tetanic seizures** in such patients, which can be misinterpreted as epileptic seizures.