"Early Morning Seizures Unmasking a Rare Metabolic Disorder: A Case of Glycogen Storage Disease Type IXc with Novel PHKG2 Mutation"

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Aims and materials

- To report an unusual cause of seizure in a 2 year old child
- To highlight glycogen storage disease type IX (GSD IX) due to PHKG2 mutation as a rare etiology
- To emphasize importance of considering metabolic disorders in children with seizures, growth delay , and organomegaly.
- 2 year old male child born out of consanguineous marriage
- Delivered vaginally, uneventful perinatal period
- Presented with GTCS from 5-6 months of age lasting <2min
- Developmental delay: attained motor milestones late

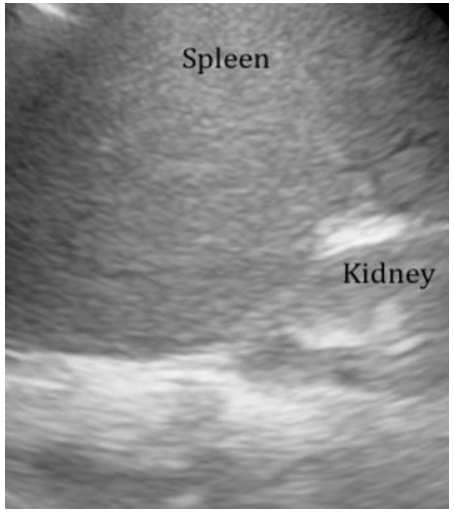
Methods (clinical features and investigation)

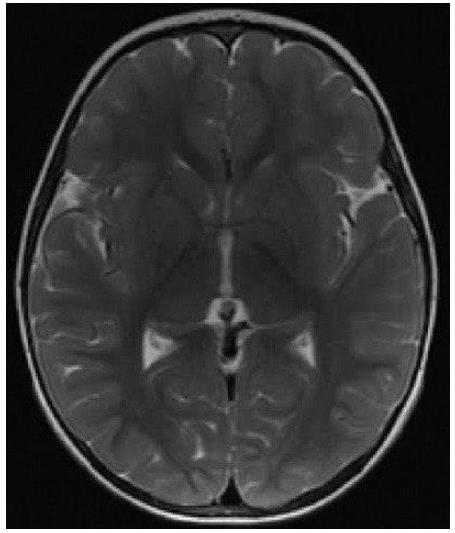
Clinical features

- Clinical examination doll like facies, hypotonia of limbs , enlarged liver and spleen.

Investigations

- Documented hypoglycemia during seizure, lab test: mild elevation of transaminases, Hb electrophoresis normal, MRI brain normal, USG splenomegaly
- Genetic testing revealed homozygous mutation in PHKG2 gene (chromosome 16p), consistent with GSD IX C.





Results and conclusion

- Child diagnosed with GSD IXC
- Clinical triad: growth delay, hypotonia, organomegaly
- Seizures associated with prolonged fasting and hypoglycemia
- GSD IX should be suspected in children with growth delay, organomegaly, and seizures linked to fasting.
- Early recognition and genetic testing are crucial for diagnosis

THANK YOU