

Severe neonatal seizures in familial hypomagnesemia with hypocalcemia: Long-term outcomes of two siblings after 12 and 23 years of treatment

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Objective: Familial hypomagnesemia with secondary hypocalcemia is a rare autosomal recessive disease caused by mutation in a gene located on 9 q 22 called TRPM 6 gene. We report the long-term outcomes of two siblings with this disease to emphasize the importance of the recognition of this illness.

Methods: The medical records of 2 siblings with familial hypomagnesemia with secondary hypocalcemia, who had been followed at the Department of Pediatrics, Ramathibodi Hospital since 1981 and 1992, were reviewed for clinical course with emphasis on mental and physical developments.

Results: The two siblings were from a family of five daughters presented with intractable seizures. The onsets were at 19 days and 28 days of age. In both, hypocalcemia was initially detected and treated with calcium and phenobarbital. Seizures were not controlled owing to unrecognized hypomagnesemia for 7 days and 5 days after the onset. After restoring the concentration of serum magnesium close to the normal levels, the seizures were completely controlled. Investigations revealed normal functions of kidneys and parathyroid. The urinary excretion of magnesium and calcium were normal. No evidence of malabsorption of other elements of nutrients from gastrointestinal tract was found. We assumed from the result of investigations that the most possible cause of hypomagnesemia of these two patients were selective defect in the intestinal absorption of magnesium. In the early stage of follow-up, both patients had occasional tonic seizures with significantly low serum magnesium levels. But soon after the daily dosages of magnesium were increased to 1 and 1.5 mg/Kg of the body weight in both siblings, no further clinical seizures occurred, and the normal levels of serum magnesium were maintained. Both patients were followed up regularly for 23 and 12 years respectively. Up to present both patients have normal physical and intellectual developments.

Conclusion: Early recognition and appropriate magnesium supplementation with good compliance are important factors in achieving the excellent outcomes of two patients with this disease.

References

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