A Rare Cause of Familial Hypomagnesemia: A Case with Trpm6 Mutation

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INTRODUCTION

- Familial hypomagnesemia with secondary hypocalcemia is a rare autosomal recessive disease characterized by very low serum magnesium levels.
- It is caused by mutations in the gene encoding the transient receptor potential melastatin 6 (TRPM6).
- It typically occurs in the first months of life with symptoms of increased neuromuscular excitability such as convulsions, muscle spasms and tetany.
- Prolonged untreated hypomagnesemia may lead to developmental delay, intellectual disability, failure to thrive and severe cardiomyopathy.
- Herein, we present a case who exhibited hypocalcemic seizure due to hypomagnesemia and was found to have a mutation in the TRPM6 gene.

CASE REPORT

- 3 months, F
- She was admitted to an external center at the age of 35 days due to contraction in the right arm and right leg.
- Magnesium and calcium treatment was started when the calcium level was 6.1 mg/dl and the magnesium level was 0.68 mg/dl in the examinations.
- In the physical examination of the patient at the age of 3 months;
  - Height: 56 cm (-1.5 SDS)
  - Weight: 6.2 kg (0.54 SDS)
  - Head circumference: 41 cm (0.81 SDS)
  - BMI: 19.7 kg / m2 (SDS: 2, 15)
  - System examinations were normal
- She was born at term, weighing 3400 g, with C/S.
- There was a first degree cousin marriage between the parents.
- The laboratory findings of the patient are presented in Table 1.
- Calcium and magnesium treatments were started.
- In the genetic analysis, c.762T> A p. (Tyr 254 *) homozygous mutation was detected in the TRPM6 gene.
- The treatment of the patient was continued with oral magnesium.
- During the follow-up, there was no recurrence and neurological development was normal.

Table 1. The laboratory findings of the patient

Calcium (mg/dL)	7,5
Magnesium (mmol/L)	0,22 (0,70-0,86)
25-OH Vitamin D (ng/mL)	62,3
Parathormone (PTH) (pg/mL)	14,5
Urine ca/kr ratio	0,17
Urinary ultrasonography	Normal

CONCLUSIONS

Although it is a rare disease, familial hypomagnesemia with secondary hypocalcemia should be considered in any pediatric patient presenting with persistent hypocalcemic seizures and severe hypomagnesemia. If diagnosed and treated early, patients can show normal physical and neurological development.

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