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CLINICAL QUIZ

An infant with recurrent convulsive seizures of 3 weeks duration: Questions

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Case summary

A 1-month-old Japanese boy was admitted to the hospital with a history of recurrent convulsive seizures of 3 weeks in duration. He was born at term (40 week gestation) at a normal birth weight of 2.76 kg following an uneventful pregnancy. He was the first child of healthy parents; there was no history of consanguinity. The infant was well until the 14th day of life when he developed convulsive seizures. On admission, the physicians observed tetanic manifestations, such as carpopedal spasms and convulsive seizures. The patient's serum calcium level was 5.0 mg/dL (normal range 8.0–10.5 mg/dL), and the inorganic phosphate level was 6.2 mg/dL (normal range 2.5–4.5 mg/dL). Calcium gluconate and parathyroid hormone (PTH) were administered intramuscularly in high doses, but the tetanic convulsions did not resolve. At that time, the serum magnesium level was measured and found to be extremely low at 0.16 mEq/L (normal range 1.44–1.81 mEq/L). The urinary excretion of magnesium was also low at 0.13 mEq/day (normal 0.31 mEq/L).

Parenteral magnesium therapy (8 mEq/day) was started. After a few days, the serum concentrations of magnesium and calcium increased to nearly normal levels, and the tetanic manifestations disappeared completely. Oral magnesium therapy (magnesium chloride, 1 mEq/kg/day) has been continued without any further tetanic manifestations. The magnesium supplement was discontinued on two occasions, and the tetanic convulsions reappeared promptly.

The fractional excretion of Mg for this patient was 2.7 % despite a very low level of serum magnesium (normal range 2.1–14.3 % in normagnesemic individuals, but is close to 0 if there is permanent depletion of magnesium), indicating the presence of a renal magnesium leak. The intestinal absorption of magnesium [(1−Mg in stool/Mg intake)×100 (%)] was lower than the amount of absorption observed in people with normal levels of magnesium (absorption in patient 19.6–32.9 %; normal levels 50.2–60.2 %). The serum calcium and magnesium levels of the parents were within normal limits.

Questions

1. What is the diagnosis for this patient?
2. What is the underlying genetic defect of this disease?
3. What is the prognosis of this disease?

The answers to these questions can be found at http://dx.doi.org/10.1007/s00467-013-2628-z.

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