An Early Spontaneous Recovery in a Hyperinsulinemia and Hypoglycemia Case with a Recently Found Mutation on the Gene ABCC8

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The most common cause of hyperinsulinemic hypoglycemia (HH) is recessive inactivating mutations on *ABCC8* and *KCNJ11*. In some patients, early (in months) or late (in years) spontaneous recovery can be seen.

In this case report, we present an early spontaneous recovery in an HH case with a new mutation on the *ABCC8* gene.

A 6-month-old girl was brought to our hospital with strabismus and arms and neck weakness. Parents are cousins. Physical examination revealed weight of 8 kg (25^{th}-50^{th} p), height 63 cm (25^{th} p), head circumference 44 cm (90^{th}-97^{th} p), and anterior fontanel of 1x1 cm (normal features); the baby was conscious, active, and the systemic signs were normal. Blood glucose was 43 mg/dL and serum insulin level during hypoglycemia was 5 µIU/mL; urine ketones were negative, cortisol, growth hormone, venous blood gas, plasma ammonia level, amino acids, carnitine, and acyl-carnitine profile were normal, thus, a diagnosis of HH was considered. 10 mg/kg/day diazoxide therapy was started and frequent enteral nutrition was planned for the case. She was discharged with normoglycemia, but two weeks later, she applied with a hypoglycemic episode. After increasing the diazoxide dose to 13 mg/kg/day and adding 5 mg/kg/day hydrochlorothiazide to the therapy, the patient was discharged with normoglycemia. A couple of weeks later, she applied with vomiting, distended abdomen, and edema. Diazoxide-associated edema was thought at first, so the diazoxide therapy was stopped and intravenous glucose infusion was applied. With frequent enteral nutrition, the blood glucose level became normal and the patient was discharged with a close blood glucose follow-up. On *ABCC8* gene's molecular analysis, there was a heterozygote c.3512delT frame shift mutation on exon 14 and a new variant (p.V679I) was detected on exon 28. It was found that the c.3512de1T mutation was from the father and p.V679I variant was from the mother. Regarding these findings, "recessively inherited hyperinsulinemia" diagnosis could not be confirmed. Focal hyperinsulinemia was questioned with 18F-DOPA PET/CT. However, when the patient was 6 months old, the diazoxide therapy was ended following the molecular diagnosis and she did not show any hypoglycemia episode at her 1.5-year-old follow-up visit. Thus, the case was assessed as spontaneous recovery.

In this report, a case with a new mutation on *ABCC8* gene causing HH was presented in order to emphasize that early spontaneous recovery can be seen in HH patients.

Key words: Hypoglycemia, hyperinsulinemia, seizure, *ABCC8*, spontaneous recovery