HADH Mutation is a Rare Cause of Hyperinsulinaemic Hypoglycaemia

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Introduction

Hydroxyacyl CoA dehydrogenase is coded by *HADH* gene. Mutations in this gene are a rare cause of recessively inherited hyperinsulinaemic hypoglycaemia (HH).

Case Report

Our first patient was diagnosed with hyperinsulinemia at the age of 40 days. When she was admitted firstly to the emergency department with seizure and her blood glucose and insulin levels were detected as 10 mg/dL and 10 mIU/mL, respectively. Ammonia level was 230 µg/dL. She was diagnosed with HH. Diazoxide therapy was began with good response to treatment. Her birth weight was 3500 g and no family consanguinity was reported. When she was 4.5 years of age, her brother was admitted to our emergency with seizure at 40 days and hypoglycemia was detected like in his sister. Blood glucose was 34 mg/dL and simultaneous insulin level was 25 mIU/mL and ammonia level was 108 µg/dL. His birth weight was 3300 g. Diazoxide was successful treatment for him as well.

Results

Sequencing analysis of the *KCNJ11*, *ABCC8* and *GLUD1* genes has failed to identify a mutation in the two siblings, whom have been diagnosed with hyperinsulinism. Sequence analysis has identified a homozygous mutation in *HADH* gene in both; they are homozygous for the *HADH* intronic mutation, c636+471G>T. This result confirms a diagnosis of recessively inherited hyperinsulinism due to a homozygous *HADH* mutation. Their parents are both heterozygous for the intronic mutation and are therefore carriers of hyperinsulinism.

Conclusion

Genetic analysis of *HADH* gene is recommended in patients with diazoxide-responsive HH from consanguineous families, who are negative for mutations in the K_{ATP} channels.