**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<sub>ATP</sub> channels.