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A Case Report of a Patient with Nonketotic Hyperglycemia Caused by a Novel Splicing Mutation in the *AMT* Gene

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KEYWORDS Alternative Splicing. *AMT Gene*. Autosomal Recessive Inheritance. Nonketotic Hyperglycinemia. Whole Exome Sequencing

ABSTRACT Nonketotic hyperglycinemia (NKH) is an inborn disease. Mutations in related genes lead to impaired glycine metabolism, allowing the accumulation of excessive glycine in body tissues. Patients usually develop severe neurological symptoms early. In this report, whole exome sequencing (WES) was applied to a female proband with hyperglycinemia and her unaffected parents. Meanwhile, cDNA sequencing was conducted to study the impact of the mutation on transcription. The results revealed that the proband exhibited a homozygous mutation of c.471+3A>G in *AMT*. The cDNA sequencing results demonstrated that this mutation resulted in the skipping of exon 4, leading to the loss of 132 nucleotides. This report enriched the study of *AMT* gene mutations. In addition, the researchers discussed the genetic and clinical heterogeneity of NKH, providing valuable insights for genetic counselling.