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GLUD 1 Mutation Causing Non Ketotic Hypoglycemia with Concomitant Hyperammonaemia: A Case Report

https://doi.org/10.15605/jafes.034.S108

Noor Arliena MA, Jeanne Wong SL, Nalini MS, Rashdan Zaki M, Pian T, Annie Leong, Teoh ST, Janet Hong YH, Fuziah MZ
Hospital Putrajaya, Putrajaya, Malaysia

INTRODUCTION

Hypoglycemia is a medical emergency which carries serious short term and long-term consequences. The time of crisis is the best time to collect critical samplings to make the diagnosis. In hyperinsulinaemic hypoglycemia (HH) due to the inhibitory effect of insulin on lipolysis and ketogenesis, there is suppressed ketone body formation, leading to increased risk of hypoglycemic brain injury. Mutations in 12 different key genes (ABCC8, KCNJ11, GLUD1, GCK, HADH, SLC16A1, UCP2, HNF4A, HNF1A, HK1, PGM1 and PMM2) that are involved in the regulation of pancreatic beta cells have been described to be responsible for the underlying molecular mechanism leading to congenital HH.

CASE

A 10-year-old girl, presented at 4 months old with afebrile seizures. Noted recurrent hypoglycemia needing glucose infusion up to 15 mg/kg/min. Born premature at 34 weeks, birthweight 2.2 kg with no postnatal complications. Physical examinations were unremarkable. Critical sampling done during hypoglycemia; RBS: 1.8 mmol/L, insulin 5.67 uIU/mL (<1), C-peptide 632 ng/mL (0.9-7.1), lactate 6.57 mmol/L (0.63-2.4), ammonia 266.9 umol/L (14.7-55.3), growth hormone 11.3 ng/ml (>10), cortisol 351 nmol/L (>250). Normal LFT and VBG. She was started on oral Diazoxide and hydrochlorothiazide and responded well. Genetic study confirmed heterozygous for a novel missense mutation, G446V, in exon 12 of the GLUD1 gene. Trial of stopping Diazoxide at 9-year-old failed.

Hyperinsulinaemia-hyperammonaemia (HI/HA) is the second most common cause of hyperinsulinaemic hypoglycemia. It is caused by mutation in GLUD1 gene resulting in a decreased inhibitory effect of guanosine triphosphate on the glutamate dehydrogenase (GDH) enzyme. HI/HA syndrome patients are Diazoxide responsive and in some cases dietary protein restriction might be necessary.

CONCLUSION

The importance of establishing the correct diagnosis in hyperinsulinism from critical samplings results and genetic study is of importance to predict the prognosis and proper counselling to patient and family.

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Graves' Disease: Clinical Features and Short-Term Outcomes

https://doi.org/10.15605/jafes.034.S109

Shiau Hooi W, Song Hai L
Department of Paediatrics, Sabah Women and Children's Hospital, Malaysia

INTRODUCTION

Graves' disease is an uncommon disease in childhood with prevalence of 0.02%. It accounts for most of the thyrotoxicosis in paediatric population. Treatment option is limited and the reported remission rate is low.

METHODOLOGY

This is a descriptive study. All patients with Graves' disease who attended the endocrine clinic in Sabah Women and Children's Hospital are enrolled. Data was obtained through review of their medical records. Their clinical features and treatment outcome were described. Results are expressed as numerical values (percentages) for categorical variables and medians (25th, 75th percentiles) for continuous variables.

RESULTS

A total of nine patients were studied whereby 78% of them are female. Their median age of presentation is 9.0 years old (3.5, 11.6). Most common presenting features are goitre, exophthalmos and tachycardia. At presentation, their median FT4 is 51 pmol/L (29, 75). Most patient had a positive thyroid receptor antibody. All patients were treated with carbimazole, median dose of 0.6 mg/kg/d (0.5, 1.0). One patient had additional thyroxine to the treatment (block and replace) due to wide fluctuation in thyroid function. None of the patients experienced side effects from treatment. Median duration of follow up is 3.4 year (1.1, 8.2). Only one patient (11%) in this cohort achieved remission.

CONCLUSION

Our study shows that achieving remission is challenging in paediatric Graves' disease. Carbimazole is a safe treatment option within the duration of follow up in our cohort.