# A Case of Hyperinsulinism/Hyperammonemia Syndrome

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## OBJECTIVES

The hyperinsulinism/hyperammonemia (HI/HA) syndrome is the second most common cause of hyperinsulinemic hypoglycemia of infancy. HI/HA syndrome is caused by a mutation in the GLUD1 gene, a gene located on chromosome 10q23.3., composed of 13 exons that encode the mitochondrial enzyme glutamate dehydrogenase (GDH). It is characterized by hyperinsulinemic hypoglycemia accompanying hyperammonemia. We report HI/HA syndrome with a 4-month-old male who hypoglycemic seizure.

## METHODS

A 4-month-old male infant presented with generalized tonic-clonic seizure. He had no symptoms of hypoglycemia when he had a generalized tonic-clonic seizure after an overnight fast. He was born after an uncomplicated $38^{+6}$-week pregnancy with birth weight of 3270 g and height of 50.0 cm. His height and weight were 67.1 cm (75 percentile) and 7.70 kg (50 percentile). His development was normal. In familial history, his parents and grandparents had no symptoms of hypoglycemia. He was suspected as having epilepsy, although no abnormality was noted in the electroencephalogram.

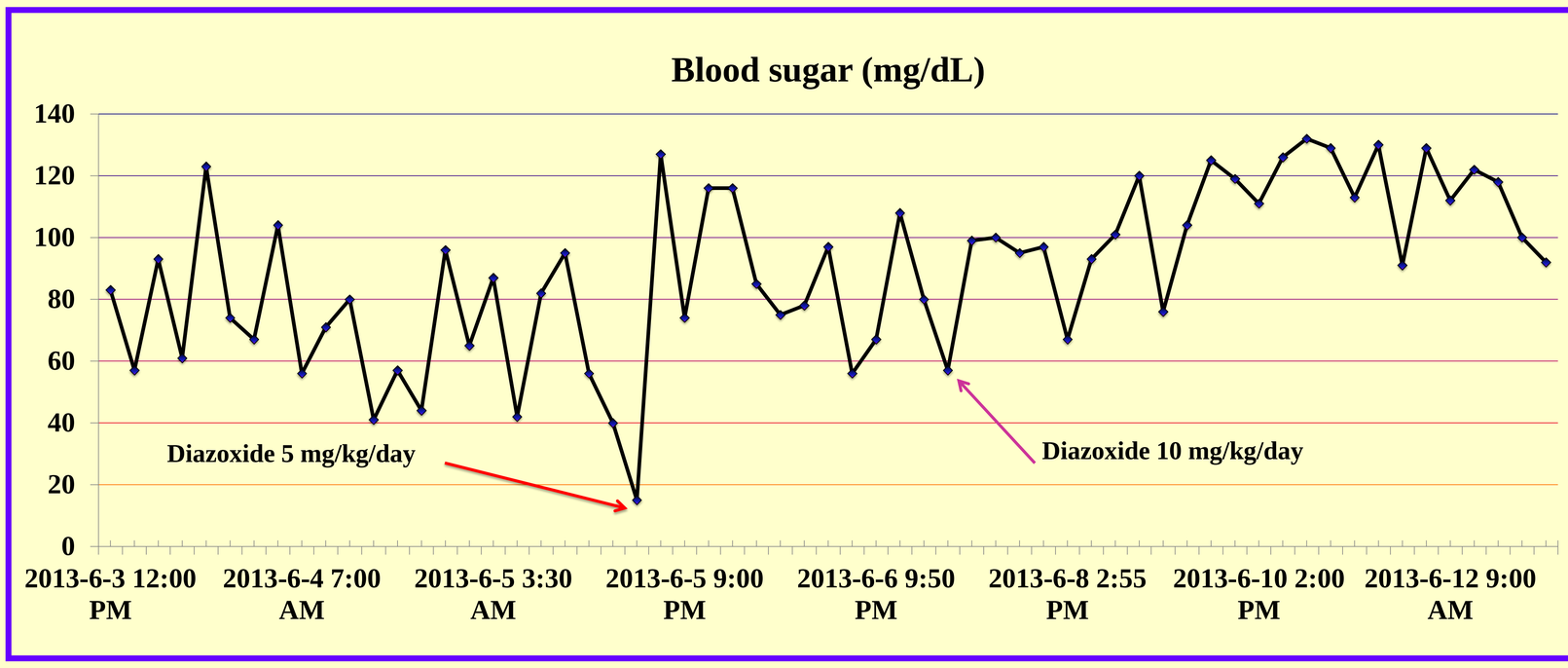


**Table 1. Laboratory finding of HI/HA patients**

| | |
|---|---|
| **HbA1c** | |
| 13-06-03 | <4.3% |
| **Insulin** | |
| 13-06-03 | 7.1 μIU/mL |
| 13-06-05 | 7.6 μIU/mL |
| **C-peptide** | |
| 13-06-03 | 2.54 ng/mL |
| 13-06-05 | 3.12 ng/mL |
| **Ammonia** | |
| 13-06-03 | 216 μmol/L |
| 13-06-05 | 169 μmol/L |

## RESULTS

At the time of seizure, the serum glucose, ionized calcium, ammonia and insulin levels were 49 mg/dL, 5.3 mg/dL, 216 μmol/L (normal <35 μmol/L) and 7.1 μ IU/mL (I:G ratio 0.15), respectively. Even though he was administered 5% dextrose fluid and was fed as usual, his blood glucose level reduced to below 50 mg/dL. He was thought to have a HI/HA syndrome. He was administered diazoxide (5 mg/kg/day), following which his blood glucose levels were not maintained within the normal range. The dose of diazoxide was adjusted by 10 mg/kg/day and leucine-free formula was applied. His blood glucose levels were maintained within the normal range.

## CONCLUSIONS

Because HI/HA syndrome is a diazoxide-responsive form of CHI, early detection and appropriate management are important to prevent brain injury. Since patients with HI/HA syndrome may have neurological complications such as developmental delay, and cognitive impairment, careful and repeated neurologic evaluation is needed.

## References

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