Hepatic glycogen synthase deficiency associated with growth hormone deficiency: A case report

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INTRODUCTION

Type 0 glycogen storage disease (GSD0) is caused by deficiency of the hepatic isoform of glycogen synthase. Growth hormone deficiency in this disorder has not been reported.

CASE

A two-year old girl who had suffered from occasional morning convulsions was admitted to our clinic. Her length and body weight were measured as 80 cm (-2.4 SD) and 11.3 kg (-1.3 SD), respectively. Physical examination was unremarkable. Metabolic profile showed fasting hypoglycaemia, hypertriglyceridemia, hyperketonaemia, hyperlactatemia, and growth hormone deficiency. Hyperglycaemia, hypertriglyceridemia and hyperlactatemia were determined after meals and in an oral glucose tolerance test. GYS2 gene analysis revealed a homozygous mutation (c.1145 G>A (p.Gly382Glu) (p.G3822E). The patient was treated human growth hormone (hGH) and uncooked corn-starch. Her height growth rate after hGH therapy was calculated as 7 cm/yr.

CONCLUSION

This is the first case with hepatic glycogen synthase deficiency associated with growth hormone deficiency. Serum insulin, c-peptid, and counter-insulin hormones at the moment of hypoglycemia were examined.