Posters

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Hyperinsulinemic Hypoglycemia and Growth Hormone Deficiency secondary to 20p11 deletion

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PMON313

Hyperinsulinemic Hypoglycemia and Growth Hormone Deficiency secondary to 20p11 deletion

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[3]Potential conflict of interest. Refer to Meeting App.

Introduction

- Hyperinsulinism (HI) and growth hormone deficiency (GHD) are causes of hypoglycemia but not commonly found together.
- A 4-month-old boy with hypoglycemia was found to have HI and GHD with subsequent genetic diagnosis of 20p11 deletion.
- 20p11 deletion has been associated with hypoglycemia due to GHD, but HI is not a common feature[1].
- Treatment with both recombinant human growth hormone (rhGH) and Diazoxide led to blood glucose (BG) normalization.

Clinical Case

- 4-month-old boy, born full-term, large for gestational age to a nondiabetic mother, presented with seizures.
- He had transient hypoglycemia at birth, normal newborn screen, and was exclusively breastfeeding with normal growth and development.
- 60-hour video EEG was normal.
- Further work-up was significant for BG of 54 mg/dL despite blood draw being 20 minutes after a breastfeed.
- Examination showed no dysmorphism, normal genitalia and neurologic exam.

Investigation

- Serum BG was 39 mg/dL while asymptomatic. Critical sample (Table 1) was significant for elevated insulin, low BOHB and low GH.
- A glucagon challenge showed an increase in BG of 26 mg/dL (35-->61 mg/dL).
- He had GH stimulation test with arginine (peak GH 3.8 ng/mL) and glucagon (peak GH 6.6 ng/mL).
- Plasma amino acid, plasma acylcarnitine profile, urine organic acids were normal.
- Brain MRI was normal.

Table 1. Results of Critical Sample

Test	Result	Normal range during hypoglycemia
Serum/plasma glucose	39 mg/dL	< 50 mg/dL
Insulin	3.2 mcIU/mL	<2 mcIU/mL
C-peptide	0.6 ng/mL	< 0.6 ng/mL
Beta-hydroxybutyrate quant (BHB)	145.6 mcmol/L	> 600 mcmol/L
Human growth hormone (GH)	4.2 ng/mL	> 10 ng/mL
Cortisol	15.7 mcg/dL	> 18 mcg/dL
Free fatty acids	0.49 mmol/L	< 0.5 mmol/L
Lactic acid	1.0 mmol/L	0.7 to 2.1 mmol/L

Treatment

- He was placed on 10% dextrose fluids with glucose infusion rate (GIR) of 8 to 11 mg/kg/min plus breastmilk.
- After starting rhGH (0.27 mg/kg/week), GIR was decreased to 3-4 mg/kg/min but he then had hypoglycemia (44-58 mg/dL).
- Diazoxide (5 mg/kg/day), and chlorothiazide (10 mg/kg/day) were started.
- Diazoxide was dose adjusted over the next days of hospitalization, until dextrose infusion was discontinued while normoglycemic.
- He was discharged on Diazoxide (12 mg/kg/day), chlorothiazide, and rhGH.
- Genetic testing with next-generation sequencing revealed 20p11.22-p11.21 deletion.

Learning Points

- Seizure can be a presenting symptom of hypoglycemia so BG should be checked in children with seizures.
- Hypoglycemia is often the result of a single etiology, but in rare instances, it can have multiple causes.
- 20p11 deletion is associated with both growth hormone deficiency and hyperinsulinism as causes of hypoglycemia.

References: Kale, T., R. Patil, and R. Pandit, A Newborn with Panhypopituitarism and Seizures. Case Rep Genet, 2017

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