PITUITARY STALK INTERRUPTION SYNDROME: a case of an infant

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BACKGROUND

PSIS

- Interrupted pituitary stalk
- Anterior pituitary hypoplasia/aplasia
- Absent/ectopic posterior pituitary

HISTORY: He was born in term via C/S. Birth weight was 3300 gr. He had been followed up for hypoglicemia and jaundice in newborn period and had no known disorder. There was no similar disorder in his family or consanguinity between parents.

CASE

PHYSICAL EXAM

- Chronological Age: 5.5 months
- Weight: 7740gr (25-50p)(-0.39 SD)
- Height: 67.5cm (25-50p)(-0.22 SD)
- SPL: 2.5 cm (<10p)
- Tanner stage: 1

SYSTEM EXAMINATIONS WERE NORMAL

- Laboratory
  - Glucose: 87 mg/dl
  - Na: 139 mEq/L
  - K: 5.6 mEq/L
  - Urine density: 1025
  - ACTH: 11.5 pg/ml
  - Cortisol: 1.27 mcg/dl
  - TSH: 8.86 mIU/mL
  - FT4: 0.76 ng/dl
  - FT3: 3.45 pg/ml
  - LH: 1.5 mIU/mL
  - FSH: 0.9 mIU/mL

- Pituitary gland MRI

- Cranial MRI: Normal

Clinical course: Low dose ACTH stimulating test found to be concordant to central adrenal insufficiency, so as TRH test to hypothalamic deficiency. Hydrocortison, L-thyroxin and Dehydrotestoron gel treatment started. In the last control, age: 1 year, Weight: 9.3 kg(25p)(-0.8 SD), Height: 75 cm (25-50p)(-0.5 SD), SPL: 4.5 cm, Puberty Tanner Stage 1, growth rate: 7.5 cm/7 month. In laboratory, glucose, plasma electrolytes were normal, and patient was euthyroid. Patient who use to receive hydrocortison 15/mg/m2/day, L-thyroxin 2.7mcg/kg/day is planned to undergo growth hormone stimulating test when the growing slow down.

DISCUSSION

- Male/Female 2.3-6.9/1
- Male age of diagnosis 9.4 11.6 years
- Heterogeneous clinical findings, most frequent is short stature (85.5%)
- 100% GH, 95.8% gonadotrophins, 81.8% corticotropin, 76.3% thyrotropin deficiency
- Hyperprolactinemia 36.4%
- Pathogenesis ?
- Birth trauma, neonatal hypokseemia
- HESX1, LHX4, SOX3, OTX2, PROKR2, GPR161 mutations

CONCLUSION

The early diagnosis of anterior pituitary deficiency is important to avoid from possible mortality and morbidity.