



An Early Diagnosis of Prohormone Convertase Deficiency

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INTRODUCTION

Prohormone convertases are a family of proteins that mediate the processing and activation of peptide hormone and neuropeptide precursors. Homozygous mutations of the PCSK1 gene result in prohormone convertase 1 deficiency characterized by diarrhea, hypoglycemia, multiple pituitary hormone deficiency beginning from neonatal period and obesity beginning after infancy.

CASE

35 days, male, referral to our hospital with hypoglycemia and metabolic acidosis

Birth/past History	Family History	Physical Examination	Laboratory	Follow up
<p>Born 3400 grams at term with APGAR 9/10</p> <p>↓</p> <p>Received phototherapy with indirect hyperbilirubinemia at the age of 3 days</p> <p>↓</p> <p>Readmitted with deterioration in general condition, dehydration, hypoglycemia, metabolic acidosis and hyperbilirubinemia at the age of 13 days</p> <p>↓</p> <p>Recovered after blood exchange, fluid and inotrop therapy but metabolic acidosis and hypoglycemia repeated after feeding</p>	<ul style="list-style-type: none"> G:1 P:1 L:1 Consanguinity + (4th degree) Cousin of mother with PCK1 gene defect (excitus at 18 months) 	<ul style="list-style-type: none"> Body weight: 3280 gr, -1,68 SD Length: 51 cm, -1,18 SD Head C.: 35,5 cm, -1,5 SD Hypotonic 	<p>Hemogram Serum electrolytes Kidney/liver function TSH/free T4</p> <p>} N</p> <p>Gonadotropins showing minipuberty</p> <p>Serum cortisol ↓ Basal: 3 µg/dl 1 mcg ACTH stim. test: 14,8 µg/dl</p>	<ul style="list-style-type: none"> 10 mg/m² hydrocortisone was initiated Diarrhea began after the first month Diarrhea stopped with Basic-CH formula and pancreatin treatment

- ❖ Central adrenal insufficiency
- ❖ Hypoglycemia
- ❖ Diarrhea
- ❖ Consanguinity
- ❖ Positive family history



Prohormone convertase deficiency?



PCSK1 gene sequence analysis

A homozygous likely pathogenic variant

NM_000439.5 c:635G>A (p.Cys212Tyr)(homozygous)

CONCLUSIONS

In the presence of hypoglycemia and metabolic acidosis in the newborn and infant period; in case diarrhea is added; prohormone convertase deficiency should be considered in the differential diagnosis.

CONTACT INFORMATION

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