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 neoantal period and obesity beginning after infancy.

INTRODUCTION

CASE

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

<table>
<thead>
<tr>
<th>Birth/past History</th>
<th>Family History</th>
<th>Physical Examination</th>
<th>Laboratory</th>
<th>Follow up</th>
</tr>
</thead>
<tbody>
<tr>
<td>Born 3400 grams at term with APGAR 9/10</td>
<td>G:1 P:1 L:1</td>
<td>Body weight:3280 gr., -1,68 SD</td>
<td>Hemogram</td>
<td>10 mg/m2 hydrocortisone was initiated</td>
</tr>
<tr>
<td>Received phototherapy with indirect hyperbilirubinemia at the age of 3 days</td>
<td>Consanguinity + (4th degree)</td>
<td>Length:51 cm., -1,18 SD</td>
<td>Serum electrolytes Kidney/liver function TSH/free T4</td>
<td>Diarrhea began after the first month</td>
</tr>
<tr>
<td>Readmitted with deterioration in general condition, dehydration, hypoglycemia, metabolic acidosis and hyperbilirubinemia at the age of 13 days</td>
<td>Cousin of mother with PCK1 gene defect (excitus at 18 months)</td>
<td>Head C.:35,5 cm., -1,5 SD</td>
<td>Gonadotropins showing minipuberty</td>
<td>Diarrhea stopped with Basic-CH formula and pancreatin treatment</td>
</tr>
<tr>
<td>Recovered after blood exchange, fluid and inotropic therapy but metabolic acidosis and hypoglycemia repeated after feeding</td>
<td>Consanguinity</td>
<td>Hypotonic</td>
<td>Serum cortisol ↓</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Positive family history</td>
<td></td>
<td>Basal: 3 µg/dl</td>
<td>1 mcg ACTH stim. test: 14,8 µg/dl</td>
</tr>
</tbody>
</table>

❖ 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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