A New Case Report of Central cortisol deficiency (isolated ACTH deficiency) in a child.

Fawzia Alyafie1, Ashraf Soliman 2#

Departments of Pediatrics - Hamad Medical Center, Doha, Qatar 1, and Alexandria University Children’s Hospital, Alexandria, Egypt 2.

Introduction

Corticotropin (ACTH) is one of several peptides derived from the POMC gene. Isolated ACTH deficiency is a very rare and life-threatening disorder. It can present during neonatal period and first year of life as a result of a specific loss of function genetic mutation affecting the gene for the pro-opiomelanocortin (POMC) or T-box transcription factor (TPIT) mutation. Rarely it may present during childhood due to unknown factors. Only seven cases have been reported in the literature.

Case Study

We are reporting an 11.5 years old Qatari who presented with generalized tonic colonic seizure on the day of admission with history of fever, ear pain and discharge, fatigue, excessive sleepiness and vomiting for 4 days. He had fever (39°C) and BP = 90/50 mmHg, heart rate = 200 b/min, with progressive reduction of blood pressure. His height SDS = 1.5 and BMI = 15 kg/m2. Pubertal hair and testicular size corresponded to Tanner II. He had post-auricular tenderness suggestive of mastoiditis, with no skin or mucus membrane hyperpigmentation.

Results

Investigations revealed:
1. Hypoglycemia of (BG= 0.9 mmol/l)
2. Normal renal and hepatic functions,
3. Normal electrolytes (Na, K, Ca, PO4 and HCO3).
4. After stopping IVF, asymptomatic hypoglycemia recurred (1.7 mmol/l, 1.8 mmol/l) which was managed accordingly.
5. Cortisol level during hypoglycemia was low (3 nmol/L).
6. Other hormones were normal: IGF1 = 117 ng/ml, TSH = 4.9 mIU/L, free T4 = 13.5 pmol/L, renin = 45.7 (NL 3-66 mU/L), aldosterone = 213 (NL 111-859) pmol/L.

Discussion

This rare condition presented with severe life threatening hypoglycemia, hypotension and absence of hyperpigmentation or electrolyte abnormalities. The presence of low ACTH and cortisol during hypoglycemia as well as no cortisol response to ACTH confirmed the diagnosis. The child has been started on stress dose of hydrocortisone for three days followed by replacement dose of oral hydrocortisone (10mg/m2/day) which resulted in total resolution of his hypoglycemia.

Conclusion

This is a rare case of isolated ACTH deficiency presented with severe life threatening hypoglycemia that responded well to hydrocortisone therapy. MRI of the pituitary was normal (no cyst). The cause of this condition remains unknown.

<table>
<thead>
<tr>
<th>Time /min</th>
<th>0 min (basal)</th>
<th>30 min</th>
<th>60 min</th>
<th>90 min</th>
<th>120 min</th>
</tr>
</thead>
<tbody>
<tr>
<td>ACTH (Pg/ml)</td>
<td>8 (low)</td>
<td>————</td>
<td>————</td>
<td>————</td>
<td>————</td>
</tr>
<tr>
<td>Cortisol (nmol/L)</td>
<td>&lt;12</td>
<td>23</td>
<td>23</td>
<td>25</td>
<td>29</td>
</tr>
</tbody>
</table>

Table 1: ACTH (normal: 10 – 60 pg/mL), basal Cortisol (normal: 185 – 624 nmol/L), peak cortisol after ACTH (> 550 nmol/L).