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

Fawzia Alyafie1 , Ashraf Soliman 2#

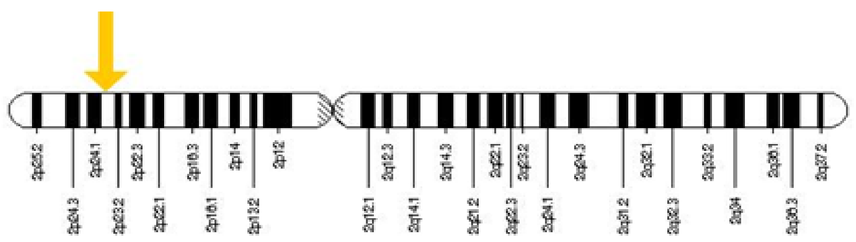
Departments of Pediatrics Hamad Medical Center, Doha, Qatar 1,
and Alexandria University Children's' Hospital, Alexandria,Egypt 2.

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 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.

Cytogenetic Location: 2p23.3

Molecular Location on chromosome 2: base pairs 25,160,852 to 25,168,870



The *POMC* gene is located on the short (p) arm of chromosome 2 at position 23.3.

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 (39C) 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 mastoditis, 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).

After stopping IVF, asymptomatic hypoglycemia recurred (1.7 mmol/l, 1.8 mmol/l) which was managed accordingly. 4. Cortisol level during hypoglycemia was low (3 nmol/L)

5. Cortisol response to ACTH (back-to-back low dose-standard dose ACTH test (table1) proved cortiol deficiency.

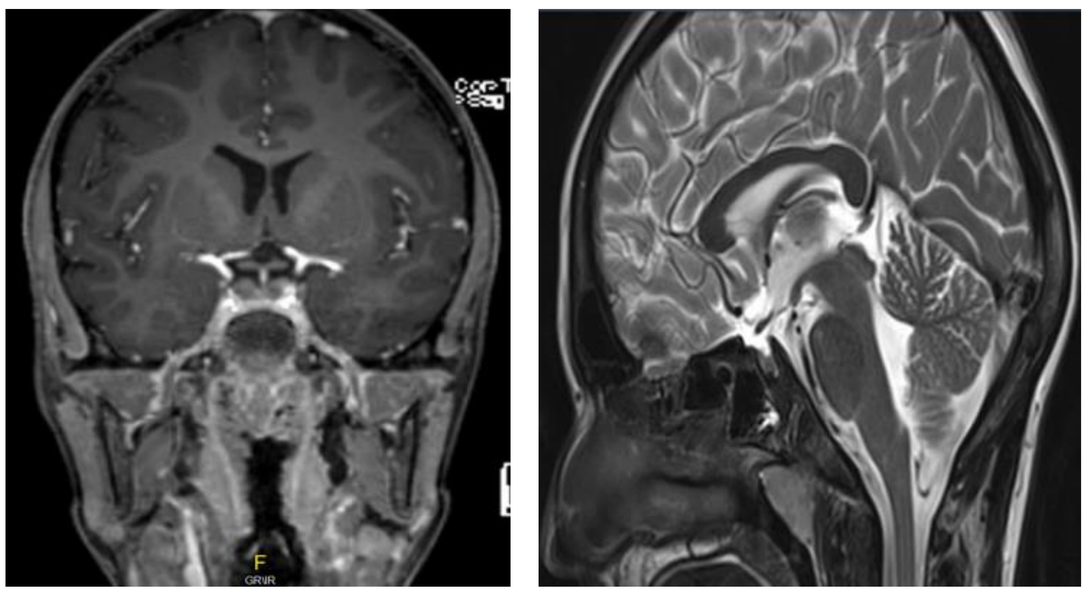
6. Other hormones were normal: IGF1 = 117 ng/ml, TSH =4.9 mIU/L, free T4 =13.5 pmol/L, renin = 45.7 (NL 3-66 mU/L), aldosterone =213 (NL (111-859) pmol/L)

ATCH stimulation test

	Low dose ACTH		Standard dose ACTH		
Time /min	0 min (basal)	30 min	60 min	90 min	120 min
ACTH Pg/ml	8 (low)	----------	----------	----------	--------
Cortisol (nmol/L)	<12	23	23	25	29

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

MRI brain



Normal MRI head

Discussion

This rare condition presented with severe life threating hypoglycemia , hypotension and absence of hyperpigmentation or electrolytes 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 threating hypoglycemia that responded well to hydrocortisone therapy. MRI of the pituitary was normal (no cyst).

The cause of this condition **remains unknown.**

ESPE 2015 601-P3

Adrenal

Fawziya Alyafei

DOI: 10.3252/pso.eu.54espe.2015

Poster presented at: 54th Annual Meeting 1-3 October ESPE 2015 BARCELONA European Society for Paediatric Endocrinology

PosterSessionOnline.com