hrp0082P2-D1-259

INADEQUATE CORTISOL RESPONSE TO TETRACOSACTIDE (Synacthen®) TEST IN NON CLASSICAL CONGENITAL ADRENAL HYPERPLASIA (NCCAH) PATIENTS: AN EXCEPTION TO THE RULE?

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Background

- ❑ **NCCAH is one of the most common autosomal recessive diseases.**
- ❑ **It may present at different ages with various degrees of hyperandrogenism.**
- ❑ **Very few studies have addressed the question of adrenal insufficiency in NCCAH patients.**

Aims of the study

To describe cortisol response to tetracosactide test in a group of NCCAH patients.

Methods

- Retrospective study, comparing cortisol response after tetracosactide test (250 µg) in NCCAH patients and a Comparison Group (CG) with precocious pubarche and 17OHP response <3 ng/mL.

- NCCAH diagnosis was confirmed by genotyping. Patients with divided into two groups: two "mild" mutations (mi/mi) or one "mild" and one "severe" mutation (se/mi).

- Adequate cortisol response was defined as a peak level >18 µg/dL.

Population description

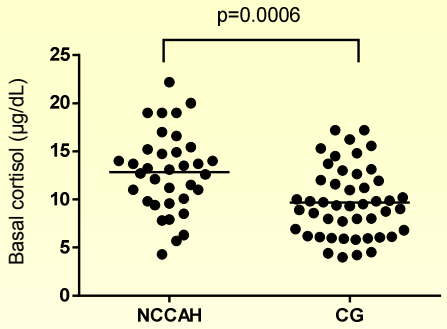
	NCCAH	CG
Girls/Boys	26/9	39/8
Age	7.0 (0.8-15.6)	7.2 (0.5-9.9)
Premature pubic and/or axillary hair	24/35 (68.6%)	47/47 (100%)
Accelerated growth rate	13/35 (37.1%)	10/47 (21.3%)
Bone age advancement (>18 months)	17/35 (48.6%)	14/47 (29.8%)
Acne	6/35 (17.1%)	10/47 (21.3%)
Fatigue	2/35 (5.7%)	0/47
Menstrual disorders	1/35 (2.9%)	0/47
Family screening	10/35 (28.6%)	NA

Conclusions

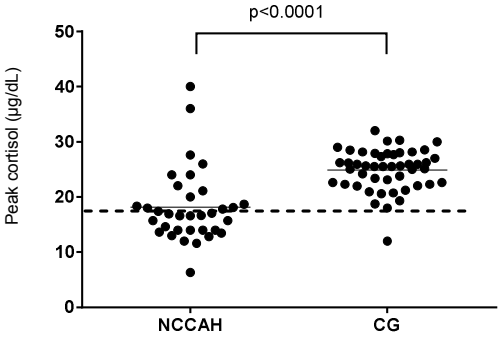
❑ Cortisol response was inadequate (<18 µg/dL) in 60% of patients with NCCAH.

❑ Hydrocortisone therapy may deserve consideration in major stress conditions (major trauma or surgery, childbirth) or objectively documented fatigue in NCCAH patients with inadequate cortisol response.

Results



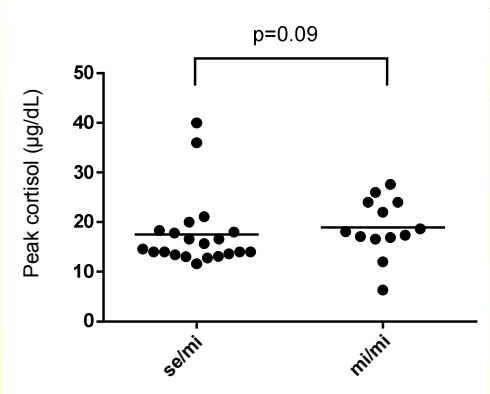
Basal cortisol in NCCAH patients: 12.9 µg/dL (4.3-22.2) Vs 9.7 (4.2-16.2) in the CG (p=0.0006).



Cortisol peak in NCCAH patients: 18.2 µg/dL (6.3-40) Vs 24.9 (12-30.3) in the CG (p<0.0001).

21/35 of NCCAH patients (60 %) had a low cortisol peak level, versus 1/47 in the CG (2.1%). Affected siblings showed similar cortisol response.

Basal ACTH was elevated in the NCCAH group: 51.3 pg/mL (9.5-160) Vs CG: 29.5 pg/mL (3.9-89.2), p=0.03.



Presence of one allele with "severe mutation" was not predictive of cortisol response.

None of the NCCAH patients had symptoms of adrenal insufficiency, but some reported fatigue which improved under hydrocortisone treatment.