Bone mineral status in adults with congenital adrenal hyperplasia due to 21-hydroxylase deficiency

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INTRODUCTION

21-Hydroxylase deficiency is the most frequent inborn error of steroidogenesis causing congenital adrenal hyperplasia (CAH). Bone status is affected by chronic glucocorticoid (GC) therapy and excess androgen exposure in patients with CAH.

Our objective is to evaluate the bone mineral metabolism and density in adulthood in a Tunisian cohort.

PATIENTS AND METHODS

This prospective descriptive study included 26 patients over 16 years of old with CAH due to 21-Hydroxylase deficiency. Their evaluation in adulthood was based on anamnestic, clinical, biochemical and radiological data.

RESULTS

-Our population is composed of 11 men and 15 women.

- **Age**: 27.4 years (16.5-48 years).
- **Phenotype**

Classic salt wasting 10 cases

Classic simple virilizing 8 cases

Non-classic 8 cases

- **Body mass index:** 26,9 ± 4,27 kg/m^2 (20,3 - 34,8 kg/m^2)
- **Glucocorticoids treatment**

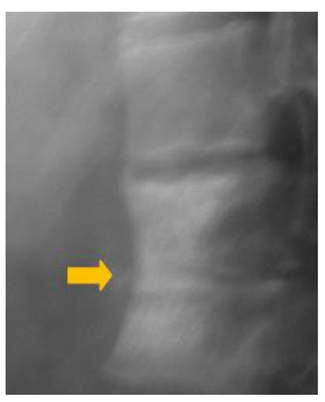
Classic form: hydrocortisone (16 cases)
41,8 ± 10,6 mg/m2/j
(28-62,8 mg/m2/j)
dexamethasone (2 cases)
0,5 mg/j
Non-classic form: hydrocortisone (5 cases)
19,8 mg/m2/j
(13-27 mg/m2/j)
dexamethasone (3 cases)
0,5-0,75 mg/j

- Biochemical and hormonal data of mineral bone status in the total population and in the different clinical forms of CAH are noted in the table. The mean serum testosterone was 6,4 ng/ml (0,8-6,7 ng/ml) and 5,9 ng/ml (1,1-8,9 ng/ml) in men and women respectively.

Table : Bone mineral status in different clinical forms of 21-hydroxylase deficiency

	Population	Classic salt wasting	Classic simple virilizing	Non-classic
Calcium (mmol/l)	2,32 ± 0,16	2,35 ± 0,14	2,33 ± 0,23	2,28 ± 0,15
Phosphorus (mmol/l)	1,08 ± 0,15	1,09 ± 0,25	1,10 ± 0,11	1,07 ± 0,12
Magnesium (mmol/l)	0,75 ± 0,11	0,79 ± 0,15	0,72 ± 0,1	0,75 ± 0,1
Albumin (g/l)	41,2 ± 1,95	41,6 ± 2,1	40,8 ± 1,8	41,1 ± 2
Alkaline phosphatase (UI/l)	160,6 ± 53,8	174,5 ± 51,3	182,3 ± 69,4	121,28 ± 40,6
Osteocalcin (ng/ml)	20,98 ± 6,47	27,64 ± 9,48	17,6 ± 3,48	15,8 ± 6,36
25-OH vitamin D (ng/ml)	15,8 ± 8,6	20,23 ± 7,61	18,5 ± 7,86	7,42 ± 4,02
PTH (pg/ml)	89,8 ± 29,4	31 ± 25,46	26,95 ± 11,5	92,5 ± 48,78

Figure: Standard dorso-lumbar spine X-ray showing a anterior vertebral compression of L1.



- Vitamin D deficiency was observed in 22 cases. Only one patient 23,5 years-old with the classic salt wasting CAH form had an anterior non-traumatic vertebral compression fracture L1-L2 (figure).
- Of 25 patients studied by bone densitometry, 10 showed bone demineralization: 1 case of trabecular osteoporosis and 9 cases of osteopenia (trabecular and cortical in 5 and 4 cases, respectively).

DISCUSSION AND CONCLUSION

Management of classic CAH consists of life-long GC replacement, and often presents a challenge because adequate androgen suppression may require GC overreplacement. Nonclassic CAH patients may require GC therapy, but lifetime GC therapy is often not warranted. As such, patients with CAH are exposed to different hormonal derangements, which may affect bone status among various other metabolic and developmental factors. It seems difficult to conclude on the bone status of adult patients with 21-hydroxylase deficiency. Studies of bone mineral density (BMD) in adult CAH patients have reported conflicting findings, ranging from low BMD to normal BMD. Factors that have been associated with low BMD in CAH populations have included excess GC exposure and low adrenal androgens. Most studies are retrospective with heterogeneous population that includes limited number of patients, often under the age of 50. However, it seems that bone density is most often preserved in patients who benefit from recent treatment protocols using more physiological doses of glucocorticoids.

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