Low adrenal androgen levels in patients with and without primary adrenal insufficiency in APECED (APS1)

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INTRODUCTION

Treatment of primary adrenal insufficiency (PAI) is often challenging in patients with autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED or APS1). In addition, electrolyte levels often fluctuate in APECED patients without clear relation to levels of mineralocorticoids [1,2].

AIM

To describe adrenal steroid and electrolyte levels in patients with APECED, with or without primary adrenal insufficiency, compared to healthy adult control subjects.

METHOD

Cross-sectional study including 42 Finnish patients with APECED (27 females, 7 under 18 years of age)[3] and 68 age- and gender-matched healthy adult control subjects (43 females).
We determined serum adrenal steroids, sodium, potassium, and creatinine concentrations, as well as blood pressure in all study participants.
Mann-Whitney U-test was used to determine differences between the groups.

RESULTS

Comparing patients with PAI (n=35) to patients without PAI (n=7):

Patients with PAI had significantly lower serum cortisol, cortisone, aldosterone, DHEA, androstenedione, and cortisol-to-cortisone ratio (Figure 1). Electrolyte levels did not differ between the groups.

Comparing adult patients with PAI (n=30) to healthy controls (n=68):

Serum concentrations of all adrenal steroids, plasma potassium, and cortisol-to-cortisone ratio were significantly lower in PAI patients (Figure 1). Plasma sodium and creatinine were significantly higher in patients with PAI compared with controls.

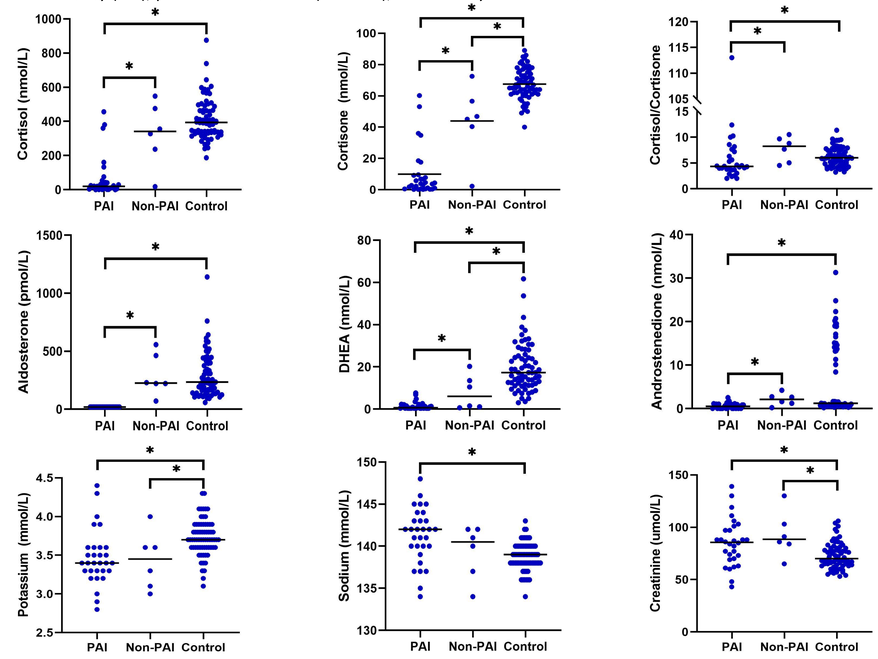
Comparing adult non-PAI patients (n=6) to healthy controls (n=68):

Concentrations of cortisone, DHEA, and potassium were significantly lower in non-PAI patients (Figure 1). Their plasma creatinine was also higher compared with controls.

Three (75%) non-PAI patients with low levels of DHEA and/or DHEAS had autoantibodies against 21-hydroxylase at the age of 23.0-62.7 years, but none had antibodies against side-chain cleavage enzyme.

Blood pressure levels did not differ between any two groups.

Figure 1. Serum adrenal steroid, electrolyte, and creatinine levels levels in adult APECED patients with primary adrenal insufficiency (PAI), patients without PAI (Non-PAI), and healthy controls. Horizontal lines indicate medians.



CONCLUSIONS

- **Adrenal steroid and electrolyte concentrations differ 1) between APECED patients with and without PAI and 2) between APECED patients with PAI and healthy controls.**
- **Concentrations of cortisone, DHEA, and plasma potassium are significantly lower in non-PAI APECED patients than in healthy controls.**
- **Low DHEA may precede the development of PAI in patients with APECED.**

REFERENCES

1 Perheentupa J. Autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy. *The Journal of Clinical Endocrinology and Metabolism 2006; 91(8): 2843-2850.*

2 Ferre EMN et al. Redefined clinical features and diagnostic criteria in autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy. *JCI Insight. 2016; 1(13): e88782.*

3 Laakso S et al. Severe phenotype of APECED (APS1) increases risk for structural bone alterations. *Frontiers in Endocrinology (Lausanne) 2020; 11: 109.*

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