PRIMARY PIGMENTED NODULAR ADRENOCORTICAL DISEASE (PPNAD) JUSTIFYING A PEDIATRIC CASE OF ACTH-INDEPENDENT CUSHING SYNDROME (CS)

Liana Capelo Costa 1, Stefânia Sporleder Vieira 1, Elisa Cordeiro Apolinário 1, Renata Fragomeni 2, Marinez Bizarro Barra 2, Cristiane Kopacek 1

1.Serviço de Endocrinologia Pediátrica - Hospital da Criança Santo Antônio - Porto Alegre - BR

2. Serviço de Patologia - Universidade Federal de Ciências da Saúde de Porto Alegre - Porto Alegre - BR

Introduction

Background: PPNAD is a very rare cause of Cushing syndrome (CS) in children and its diagnosis may be very challenging. Herein we report on a very young boy presenting with classical features of CS, including severe life-threatening blood hypertension and infections, caused by PPNAD.

Case report

A two-years-old boy presenting with excessive weight gain, acne, axillary odor, hirsutism, and behavioral and mood changes for up to nine months (Figure 1). He presented with typical signs of CS including moon face, facial acne, hirsutism, mild virilization with pubertal Tanner stage G1Ph2, and tachycardia. Blood hypertension was severe and very difficult to manage, reaching up to 160x100mmHg. Biochemical investigation (Table 1) excluded exogenous CS (basal plasma cortisol 25 mcg/dL-698nmol/L) and confirmed ACTH-independent (ACTH= 2.4pg/mL-0,53pmol/L) endogenous CS (morning plasma cortisol post-1mg DEX= 23 mcg/dL-634nmol/l). Abdominal MRI reveal slightly enlargement of both adrenals with a small isolated 5mm nodule on left side. Then, the diagnosis of PPNAD was suspected. Left adrenalectomy was performed with intraoperative frozen section examination, which revealed architectural changes suggestive of PPNAD. Following, bilateral adrenalectomy was completed and the diagnosis was confirmed by pathology (Figure 2). After the adrenalectomy, the patient received stress-covering Hydrocortisone doses, which was subsequently reduced to physiological dose. Patient's outcome was favorable, with rapid improvement of acne and tissue infiltration but the regression of hypertension took longer time (Figure 3). He was discharged under oral glucocorticoid and mineralocorticoid replacement. Initial investigation clinical, biochemical and imaging screening (skin, testis, heart, thyroid, and pituitary) did not reveal signs of Carney complex. Molecular analysis did not reveal germ line pathogenic variants in the PRKAR1A gene.

Figure 1. Clinical signs

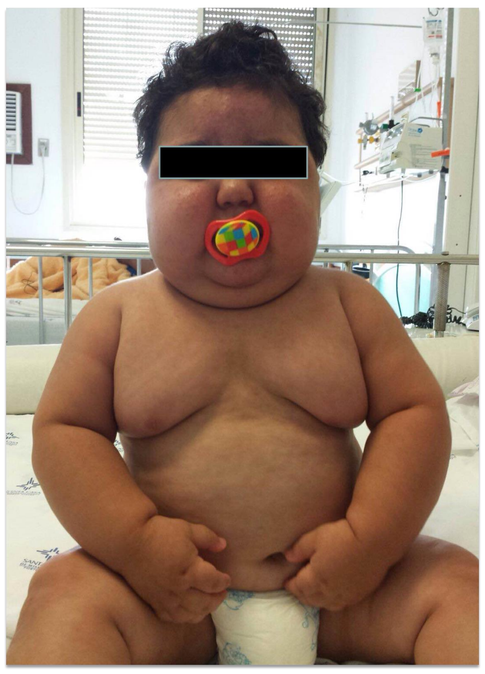

Table 1. Labor values

	Values
Creatinine	0,22 mg/dL
Urea	19 mg/dL
Na	140 mEq/L
K	4,5 mEq/L
Glucose	66 mg/dL
Cortisol 7h30	31,2 mcg/dL
Testosterone	61 ng/mL
ACTH	< 5 pg/mL
DHEA-S	46 mcg/dL
Androstenedione	3,1 ng/mL

Figure 2. Histological findings

Figure 3. Follow up

Conclusions

In children, the diagnosis of CS may be challenging, mainly in the case of PPNAD, which accounts for less than 2 % of ACTH-independent CS. In this situation, imaging exams may not show gross abnormalities and the diagnosis is only confirmed by pathology. In addition, up to 70% of the PPNAD patients and/or their families present or will develop other signs of Carney complex, a serious life-threatening condition.

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Adrenal

cristiane kopacek

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