ADDISON'S DISEASE: DELAY IN DIAGNOSIS IN A GIRL WITH LONGSTANDING SYMPTOMS

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BACKGROUND

Autoimmune destruction of the adrenal cortex is the cause of primary adrenal insufficiency (AI) in 45% to 55% of cases in children

CASE PRESENTATION

- 10 $^{10/12}$ year old girl referred because of suspicion of AI
- Longstanding complaints of extreme fatigue, loss of appetite, recurrent gastric symptoms and salt craving

MEDICAL HISTORY

2 years: Idiopathic thrombocytopenic purpura (ITP)

4 $^{11/12}$ years: Hospitalization with vomiting, dehydration and electrolyte abnormalities.

She was found underweight (BMI SDS: - 5.71), with a biochemical profile (table 1) indicative of AI (hypoglycemia, hyperkalemia, hyponatremia and inappropriately high sodium excretion). She was managed with iv hydration. The family failed to attend a scheduled appointment at the Pediatric Endocrinology Clinic.

Table 1: Biochemical profile at the age of 4 $^{11/12}$ years

Test	Result	Reference range
Glucose (mg/dl)	56	70-100
Urea (mg/dl)	39	5-45
Creatinine (mg/dl)	0.4	0.5-1.0
Potassium (mEq/L)	5.8	3.5-5.5
Sodium (mEq/L)	131	134-148
Calcium (mg/dl)	8.8	8.6-10.6
Urine sodium (mmol/l)	50	<25

At the age of 10 $^{10/12}$ years

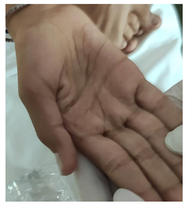
Extreme exhaustion and daily consumption of large amounts of salt prompted a second referral by the pediatrician.

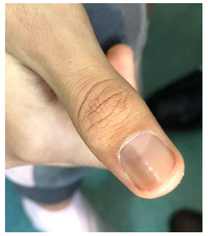
Somatometry
Height: 146 cm (67th percentile)
Weight: 25 kg (1st percentile)
BMI SDS: - 4.26 (underweight)

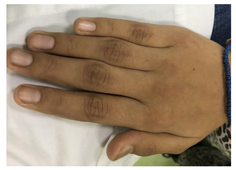
Tanner stages
Breast: Tanner II-III
Pubic hair: Tanner I
Axillary hair: Tanner I

Physical examination
normal female external genitalia
generalized hyperpigmentation
no mucosal candidiasis
blood pressure 93/66 mm Hg

Fig 1-4*:* Hyperpigmentation







Laboratory tests	Result	Reference range
cortisol (μg/dl)	0.7	6.2 – 19.4
ACTH (pg/ml)	5690	7 - 64
PRA (ng/mL/h)	24.48	0.5-4.7
aldosterone (ng/dl)	11.8	3.0-28
DHEA-S (μg/ml)	0.067	0.4-1.4
Δ4-A (ng/ml)	0.16	0.25-0.8
TSH (μIU/ml)	6.2	0.4-5.0
fT4 (ng/dl)	1.06	0.9-1.9

250 μg cosyntropin stimulation test (Synacthen®)		
	17-OHP	Cortisol
0	0.32 ng/ml	0.7 μg/dl
30 min	0.31 ng/ml	0.75 μg/dl
60 min	0.26 ng/ml	0.73 μg/dl

adrenal cell autoantibodies (ACA) = 1:20	autoimmune Addison's disease
TGAb = 203.9 IU/ml	autoimmune thyroid disease
TPOAb < 1.00	
h-tTG IgA: 107.3 (<10)	celiac disease
EMA IgA, IgG: positive	
glucose: 80 mg/dl	normal glucose and electrolytes no signs of hypoparathyroidism
Na: 135 mEq/L, K: 4.4 mEq/L	
Ca: 9.4 mg/dl, P: 5.0 mg/dl	

DIAGNOSIS AND TREATMENT

- She was started on stress doses of hydrocortisone (HC) with dramatic improvement in clinical status and then transitioned to oral HC and fludrocortisone
- Type 2 autoimmune polyglandular syndrome (**APS-2**) was diagnosed

Major manifestations
autoimmune Addison's disease
+
autoimmune thyroid disease

Minor manifestations
celiac disease
+
history of ITP

→ **APS-2**

CONCLUSIONS

Clinical and biochemical profile of the patient at the age of 4 years and 11 months elucidates that the diagnosis was present for over 6 years.

This case report highlights that symptoms of adrenal insufficiency are often underestimated by patients and possibly physicians, leading to delayed diagnosis.

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