A case of X-linked adrenoleukodystrophy presenting with primary adrenal insufficiency and normal VLCFA

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Introduction

- X-linked adrenoleukodystrophy (X-ALD) is a rare autosomal recessive neurodegenerative disease caused by a mutation in the *ABCD1* gene. Although its clinical presentation varies, X-ALD is generally characterized by progressive demyelination of the central nervous system, primer adrenal insufficiency, and elevated plasma very long-chain fatty acid (VLCFA) levels. Herein, we aimed to present a case of X-ALD with normal VLCFA caused by a pathogenic variant in *ABCD1* gene.

Case: 12 years 7 months ♂

Complaint

Hyperpigmentation,

Weakness,

Downward trend in his school performance since last year

Background and Medical histories

He was born with birth weight of 1,750 grams at 32 gestational weeks after uneventful pregnancy

His neuromotor milestones were normal.

The parents were no relatives.

Two of his cousins were followed by another center due to adrenal insufficiency.

Physical examination

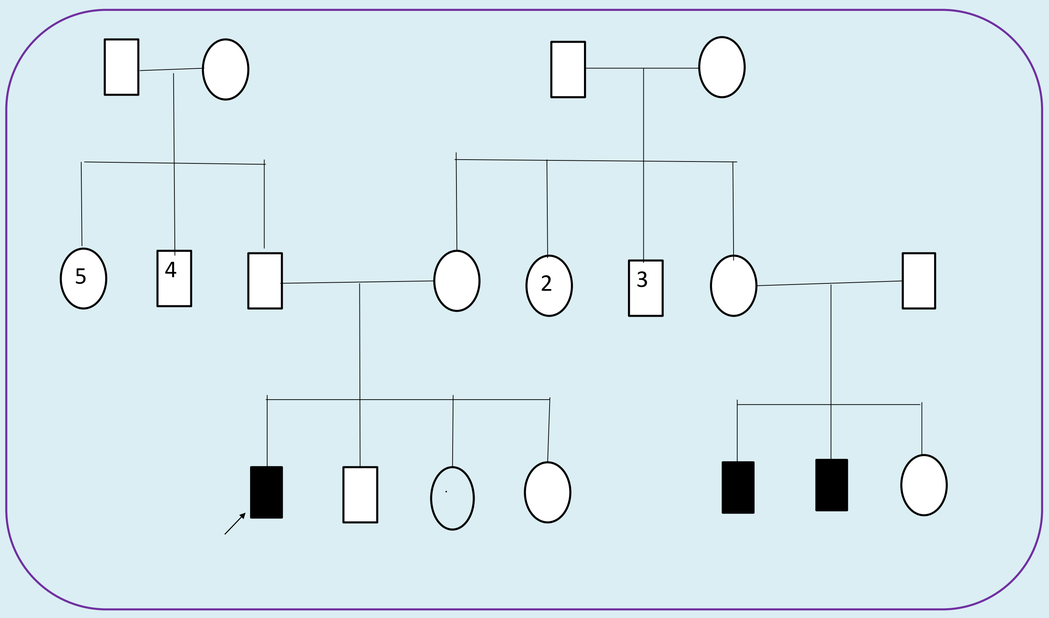
Height was 165.6 cm (1.32 SDS), weight was 46.2 kg (-0.16 SDS), BMI 16.9 kg/m2 (-0.99 SDS),

Blood pressure was 110/70 mmHg (50th centile),

His puberty was compatible with Tanner Stage 4,

Muscle strength was 5/5 and hyperpigmentation was observed in his oral mucosa and nipples

Pedigree



Laboratory examination

Parameters	Value	Normal	Parameters	Value	Normal
Hemoglobin (gr/dl)	13,2	(11-16)	Aldosterone (pg/ml)	66	(82-192)
Sodium (mmol/l)	139	(136-145)	Plasma Renin Act. (ng/ml/s)	2,01	(0,5-3,3)
Potassium (mmol/l)	4,4	(3,4-5)	TSH (µIU/ml)	1,6	(0,5-4,53)
Glucose (mg/dl)	96	(60-100)	Free T4 (ng/dl)	1,16	(0,7-1,48)
BUN (mg/dl)	12	(7-16,8)	T. cholesterol (mg/dl)	134	(0-170)
Creatinine (mg/dl)	0,6	(0,3-0,7)	Triglyceride (mg/dl)	163	(23-145,3)
ACTH (pg/ml)	>1250	(0-46)	HDL (mg/dl)	40	(35-75)
Cortisol (µg/dl)	3,8	(3,7-19,4)	LDL (mg/dl)	61	(60-130)

VLCFA

- C22:32 mg/L (N:10,5-51)
- C24:28 mg/L (N:8,5-37,5)
- C26:0,3 mg/L (N:0,1-0,6)
- C24/C22:0,83 (N:0-1,16), C26/C22:0,01 (N:0,02)

Cranial MRG

- Cranial T2A-FLAIR A MRI revealed bilateral hyperintense areas in parieto-occipital white matter.

VEP/ BAEP

- Normal

Genetic analysis of ABCD1

- A hemizygouspathogenic variant in exon 6 that was previously reported was detected [NM_000033:c.1571G>A(p.Trp524*)]

Clinical follow-up

- Hydrocortisone treatment was started for adrenal insufficiency.
- Family screening is planned.
- Recently, the patient is followed by pediatric metabolism and pediatric neurology clinics.

Conclusion : In patients with primary adrenal insufficiency, X-related adrenoleukodystrophy should be considered in differential diagnosis even if plasma VLCFA levels are normal. *ABCD1* gene analysis should be considered in the presence of clinical suspicion and radiological findings.