AN ADOLESCENT CASE WITH ADRENOLEUKODYSTROPHY DIAGNOSED AFTER DETECTION OF LEYDIG CELL DYSFUNCTION

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INTRODUCTION

X-linked adrenoleukodystrophy (X-ALD) is an inherited peroxisomal disease characterized by beta oxidation disorder that causes the accumulation of very long chain fatty acids (VLCA) in all tissues. It presents with clinical signs due to accumulation of VLCA in brain white matter, testes, adrenal cortex and skin fibroblasts (1).

AIM

The process leading to the diagnosis of X-ALD at presentation with nonprogressive puberty, adrenal insufficiency and Leydig cell dysfunction will be discussed.

METHOD

Case presentation:

- A 16-years and 4-months-old male patient applied to the outpatient clinic due to not entering puberty and the lack of deepening in his voice
- On physical examination, his height (173.3 cm; SDS: -0.06), body weight (53 kg; SDS: -1.55), body mass index (16.7 kg/m²; SDS: -1.9), testicular volume (25-25 ml), and stretched penile length (9.5 cm) were measured. His pubic hair growth was in stage-Il.
- Remarkable laboratory test results were as follows:
  - FSH: 3.34 mIU/mL (0.61-7.9), LH: 16.12 mIU/mL (0.5-8).
  - Total testosterone: 31 ng/dL (300-1200)

In his neurological examination, there was only slight inability in heel walking.

In brain magnetic resonance imaging, hyperintense signal changes in the bilateral thalamus, posterior leg of the internal capsule, posterior pons, bilateral parieto-occipital white matter and corpus callosum splenium were observed in consistent with X-ALD LOES score reported as 4.

No spinal involvement in spinal MR.

In laboratory tests on the inconsistency of the testosterone level with the testicular volume and the retardation of the pubic hair stage, ACTH was found 1014 pg/mL (223.3 pmol/L), cortisol: 3.5 μg/dL (96.5 nmol/L), DHEA-SO4: 10 μg/dL (0.27 μmol/L), and 14-androstenedione: 0.12 ng/mL (0.41 nmol/L).

In the VLCA panel,

- C24: 118.07 μmol/L (37.14-79.4),
- C26: 4.25 μmol/L (0-1.3),
- C24/C22: 1.97 (0.689-1.008),
- C26/C22: 0.07 (0.011-0.026) were higher.

IN 6TH MONTH FOLLOW-UP

In laboratory tests on the inconsistency of the testosterone level with the testicular volume and the retardation of the pubic hair stage, ACTH was found 1014 pg/mL (223.3 pmol/L), cortisol: 3.5 μg/dL (96.5 nmol/L), DHEA-SO4: 10 μg/dL (0.27 μmol/L), and 14-androstenedione: 0.12 ng/mL (0.41 nmol/L).

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IN 12TH MONTH FOLLOW-UP

In laboratory tests on the inconsistency of the testosterone level with the testicular volume and the retardation of the pubic hair stage, ACTH was found 1014 pg/mL (223.3 pmol/L), cortisol: 3.5 μg/dL (96.5 nmol/L), DHEA-SO4: 10 μg/dL (0.27 μmol/L), and 14-androstenedione: 0.12 ng/mL (0.41 nmol/L).

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CONCLUSIONS

In adolescents presenting with Leydig cell dysfunction, primary adrenal insufficiency should be screened and if primary adrenal insufficiency is detected, adrenoleukodystrophy should be investigated.

REFERENCES


CONTACT INFORMATION

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