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Case Presentation
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Non-Classical Congenital Adrenal Hyperplasia due to Partial 21-Hydroxylase Deficiency Revealed by Severe Hirsutism in Adulthood

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Abstract

Non-Classical Congenital Adrenal Hyperplasia (NCCAH) due to partial 21-hydroxylase deficiency is a frequent and often underdiagnosed cause of hyperandrogenism in women. We report the case of a 39-year-old woman presenting with severe hirsutism. Hormonal evaluation revealed elevated 17-Hydroxyprogesterone (17OHP), confirmed after ACTH stimulation, consistent with NCCAH. Imaging findings were normal. This case highlights the importance of systematic screening for this condition in the evaluation of hyperandrogenism.

Introduction

Non-Classical Congenital Adrenal Hyperplasia (NCCAH) due to partial 21-hydroxylase deficiency is an autosomal recessive disorder caused by mutations in the *CYP21A2* gene. It results in partial impairment of cortisol synthesis, leading to accumulation of steroid precursors, particularly 17-Hydroxyprogesterone (17OHP), and excess androgen production. NCCAH accounts for more than 90-95% of late-onset forms of congenital adrenal hyperplasia and represents a major differential diagnosis in women with hyperandrogenism [1, 2].

Case Presentation

A 39-year-old woman presented with progressive hirsutism. Clinical examination revealed a Ferriman–Gallwey score of 25, consistent with severe hirsutism. There were no clinical signs suggestive of Cushing syndrome, and pubertal development was normal.

Laboratory investigations showed:

- Total testosterone: 0.8 ng/mL
- Δ4-androstenedione: 5 ng/mL (elevated)
- Basal cortisol: 180 ng/mL (within normal range)
- Basal 17-hydroxyprogesterone (17OHP): 7 ng/mL

Following ACTH (Synacthen) stimulation, 17OHP increased to 15 ng/mL, confirming the diagnosis of partial 21-hydroxylase deficiency.

Pelvic ultrasound and adrenal computed tomography were unremarkable.

Discussion

NCCAH differs from the classical form, which presents in infancy with virilization and/or salt-wasting due to severe enzyme deficiency. In contrast, NCCAH results from partial enzymatic impairment and typically manifests during adolescence or adulthood [2, 3].

Hirsutism is the most common presenting symptom and may be associated with menstrual irregularities or infertility. Given its prevalence, NCCAH should be systematically considered in the etiological workup of hyperandrogenism [4].

The diagnosis is based on measurement of 17OHP, particularly after ACTH stimulation. Current guidelines define a post-stimulation 17OHP level >10 ng/mL as diagnostic [1, 5].

Imaging studies are usually normal and are mainly useful to exclude adrenal or ovarian tumors.

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Genetic confirmation through analysis of the *CYP21A2* gene is recommended, especially for genetic counseling. Family screening may also be proposed due to the autosomal recessive mode of inheritance [2, 6].

Management depends on symptom severity and reproductive goals. Glucocorticoid therapy may be indicated to reduce androgen excess, and may be combined with antiandrogen or cosmetic treatments when necessary [3].

Conclusion

NCCAH should be considered in adult women presenting with severe hirsutism. Measurement of 17OHP, particularly after ACTH stimulation, is essential for diagnosis. Appropriate management allows symptom control and enables adequate genetic counseling.

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