

Case Report

Congenital adrenal hyperplasia with polycystic ovarian disease: A rare cause of hirsutism

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Abstract Hirsutism may be defined as presence of terminal hairs in women with a male-like pattern of distribution. Hirsutism is related to serum androgen and cutaneous sensitivity to other hormones. The most prevalent causes of hirsutism are polycystic ovary syndrome and isolated hirsutism. Non-classical congenital adrenal hyperplasia (21-hydroxylase deficiency) and drug induced type are less frequent rather rare causes. Diagnostic evaluations should address identifying etiology and its associated co-morbidities to properly manage the patients. We present a case of 22-year-old unmarried female who presented with hirsutism and on detailed evaluation she was diagnosed as a case of nonclassical adrenal hyperplasia and polycystic ovaries. Along with laser hair removal, proper treatment of underlying etiology led to reversal of her symptoms.

Key words

Hirsutism, non-classical congenital adrenal hyperplasia, polycystic ovarian disease.

Introduction

Hirsutism may be defined as the presence of terminal hair in the women, with a male pattern of distribution.^{1,2} The clinical presentation is variable, from isolated hirsutism to the presence of other signs of hyperandrogenism, menstrual irregularities and/or infertility. Hirsutism is related to serum androgens and the cutaneous sensitivity to these hormones. The most prevalent causes of hirsutism are polycystic ovary syndrome and isolated hirsutism, in the presence of ovulatory cycles. Non-classical congenital adrenal hyperplasia (21-hydroxylase deficiency) and drug-induced hirsutism are less frequent causes.^{3,4} Androgen-secreting neoplasms and Cushing syndrome are rare etiologies related to hirsutism. Diagnostic evaluation should address on identifying the

etiology and potential risk for associated comorbidities. The aims of the treatment are: to suppress androgen overproduction, if present; to block androgen action on hair follicles; to identify and treat patients according to underlying pathology.

We present a case of 22-year-old female who presented with hirsutism and on detailed evaluation she was diagnosed as case of nonclassical adrenal hyperplasia and polycystic ovaries.

Case report

A 22-year-old, unmarried, female, resident of Gujranwala, presented in outpatient Department in Naseer Hospital, Lahore with severe hirsutism over chin for 5 years. She also had oligohypomenorrhea for 5 years for which she was referred to Gynecology Department, Shalamar Hospital, Lahore. She had severe hirsutism over chin (beard-like), upper lip, and sideburns which made her social life difficult.

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Figure 1 Severe hirsutism on chin and upper lip.



Figure 2 Severe hirsutism on the chin area.

She was using razor for hair removal frequently so she was referred for laser therapy. Her previous menstrual cycle was 6/28 days regular with average blood flow. Then she developed oligohypomenorrhea with menstrual cycle of 2/60-90 days and had only spotting. Her weight has been increased significantly from 66 to 74kg in last 3 years. However, she had no history of heat or cold intolerance, acne, pain abdomen, vaginal discharge, urinary or bowel complaints, headache, diplopia and drug intake especially hormonal therapy.

Her physical examination revealed height of 160cm and weight 74kg with BMI 28.9kg/m², blood pressure 130/80 mmHg. Severe hirsutism was noted on chin, upper lip, and sideburns (**Figures 1** and **2**). Hair growth on rest of body was normal. Her abdominal examination was unremarkable except obesity. Her pelvic examination showed male type of hair growth but normal looking genitalia without clitoromegaly.

Laboratory investigations revealed Hb 15g/dl, blood sugar random 100mg/dl, serum FSH 6.0mIU/ml, serum LH 13.90mIU/ml (ratio of FSH to LH reversed), serum prolactin 23.40ng/ml, serum testosterone 56 ng/dl (normal range 15-70 ng/dl), serum insulin 8.27 IU/ml, serum dehydroepiandrosterone sulphate (DEHA-S) 748.0µg/dl (normal range 35-430 µg/dl), 17-OH progesterone 310ng/dl (normal range 10-285 ng/dl).

Her ultrasonography report showed: uterus anteverted, normal size 5.7x3.3x3.6cm with normal midline echo; right ovary 2.9x2.8cm, left ovary 3.1x3.2cm, both had follicles of less than 9mm, peripherally arranged giving polycystic appearance; right adrenal gland was prominent 2.2cm, hypoechoic with regular outline; left adrenal gland was also prominent 2.1cm,

hypoechoic, homogenous texture; liver, gall bladder, spleen, both kidneys appeared normal; and no abnormal fluid or mass was seen.

So, etiology of her hirsutism was congenital adrenal hyperplasia and polycystic ovarian disease and was managed by collaboration of Dermatology and Gynecology Departments. She was given low dose prednisolone and laser therapy for CAH whereas PCO was managed by weight reduction, use of metformin and Dinette® (combination of estradiol and cyproterone acetate). Her follow up revealed marked improvement in hirsutism and menstrual cycle became regular with normal menstrual cycle.

Discussion

Congenital adrenal hyperplasia (CAH) is caused by congenital insufficiency of the enzyme 21-hydroxylase (21-OHD) in the cortisol synthesis pathway. Because of the virilizing effects of androgens overproduction, affected girls present with hirsutism, amenorrhea, clitoromegaly.⁵ The clinical characteristics of non-classical congenital adrenal hyperplasia (NC-CAH) do not differ markedly from those in patients with polycystic ovary syndrome or idiopathic hirsutism.^{6,7} NC-CAH is diagnosed by confirming 17-hydroxyprogesterone and androstenedione with low levels of cortisol.⁸ Polycystic ovarian syndrome, the most frequent cause of hirsutism,⁹ has estimated prevalence of 10% among women of reproductive age group.¹⁰ The Rotterdam criteria used to define PCOS are: a) menstrual cycle abnormalities, amenorrhea, oligomenorrhea, or long cycles; b) clinical and/or biochemical hyperandrogenism; and c) ultrasound appearance of polycystic ovaries. The presence of two of these three criteria is sufficient for diagnosis of PCOS once all other diagnoses have been ruled out.¹¹ Hence, along

with history and thorough clinical examination, laboratory investigations are essential in women with moderate to severe hirsutism to identify underlying etiology.¹

The aim of treatment of hirsutism is use of physical methods like lasers to improve the aesthetic appearance and to treat underlying pathology like NC-CAH and PCOS. Typical treatment of NC-CAH is low dose dexamethasone 0.25mg at bedtime to suppress androgen overproduction which generally reverses the hyperandrogenic signs.^{12,13} For PCOS, oral contraceptive pills with antiandrogenic activity are recommended as first line of treatment and disturbed menstrual cycle.⁴ In women with hirsutism, hyperandrogenism and insulin resistance, insulin sensitizers are effective for hirsutism as well as for hyperinsulinemia, hyperandrogenism and infertility.⁴ In women with NC-CAH and PCOS, as prevalence is 2.68%, patient should be given low dose dexamethasone and oral contraceptive pills with antiandrogenic activity and insulin sensitizer. Such patients need regular follow up especially for future fertility. Although subfertility is mild in NC-CAH,¹⁵ but may need ovulation induction with clomiphene citrate, gonadotrophins along with dexamethasone and metformin and even may require *in vitro* fertilization (IVF).¹⁶

Conclusion

Diagnostic evaluation of hirsutism should be done to identify etiology and along with using physical methods of treating hirsutism, underlying etiology should be addressed and treated properly.

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