Late-onset Congenital Adrenal Hyperplasia or Early-onset Polycystic Ovarian Syndrome: A Clinical Dilemma

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ABSTRACT

Aim: To differentiate nonclassical congenital adrenal hyperplasia (NCCAH) from polycystic ovarian syndrome (PCOS) in a 13½-year-old girl.

Introduction: Hirsutism and virilization are effects of hyperandrogenism by ovaries and adrenal glands. It has a marked psychological and social impact affecting the quality of life. About 75% of premenarchal girls have hyperandrogenism due to PCOS but late-onset CAH cannot be ruled out, and this leaves the clinician in a quandary regarding the diagnosis and management.

Case report: A 13½-year-old girl presented with excessive facial hair, hoarseness of voice, and darkening of elbow pits since past 2 months, which was increasing in severity. The patient had not yet attained menarche but had pubarche 1 year earlier. Examination revealed presence of acanthosis, underdeveloped breasts, and clitoromegaly >3 cm. Levels of 17-hydroxyprogesterone were normal but higher levels were reported poststimulation. Fasting insulin levels were also high. Appropriate treatment was started, which led to improvement in patient’s symptoms.

Conclusion: There is significant overlapping between PCOS and NCCAH, which warrants accurate diagnosis based on hormonal analysis to institute early and appropriate therapy.

Clinical significance: Early therapy can prevent infertility and androgenic complications later in life.

Keywords: Clitoromegaly, Hirsutism, Hyperandrogenism, Late-onset congenital adrenal hyperplasia, Polycystic ovarian syndrome.

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CASE REPORT

A 13½-year-old girl presented with excessive facial hair, hoarseness of voice, and darkening of elbow pits since past 2 months, which was increasing in severity. The
patient had not yet attained menarche but had pubarche 1 year earlier.

There was no history of acne, baldness, excessive weight gain, or fatigue.

There was no history of similar complaints in family. Her parents had a nonconsanguineous marriage.

Patient had normal intelligence quotient. General physical examination revealed normal blood pressure levels, with body mass index of 14.7 kg/m², breasts were underdeveloped with Tanner stage 2, there was presence of acanthosis nigricans at nape of neck and elbow folds and also excessive axillary hairs. Masculine voice was also evident. Grading of hirsutism according to Ferriman–Gallwey scoring revealed a score of 22, which suggested severe hirsutism.

Systemic and per abdomen examinations were normal.

On external genital examination excessive pubic hair (Tanner stage 4) was present, clitoromegaly of more than 3 cm was present, rest of genitalia was normal with intact hymen and normal vaginal orifice as depicted in Figure 1. On per rectal examination, small uterus could be made out.

Investigations revealed raised levels of serum testosterone (1.84 ng/mL), serum 17-hydroxyprogesterone levels of 2.8 ng/mL and ACTH stimulation 31.4 ng/mL, serum dehydroepiandrosterone sulfate was normal (1.92 ng/mL). She had normal levels of serum luteinizing hormone (LH 2.32 mIU/mL), serum follicle-stimulating hormone (FSH; 2.11 mIU/mL), serum progesterone (0.2 ng/mL), serum prolactin (18 ng/mL), serum thyroid-stimulating hormone (TSH; 2.6 mIU), and normal liver function test and kidney function test. But fasting serum insulin was raised (159 mU/L) and fasting and postmeal blood sugars were also normal. Ultrasonography (USG) of abdomen revealed no abnormality. The USG pelvis revealed bilateral bulky polycystic ovaries and normal uterus with 3 mm endometrial thickness. Barr body was positive. Karyotyping report was 46 XX.

After proper counseling of the patient and relatives, patient was prescribed tab Aldactone 25 mg TDS, tab Bigomet SR 250 mg BD gradually increased to 850 mg BD, calcium supplements 500 mg HS, Vit D3 sachet once a month, and Nervijen injections 1 mL intramuscular once a week, and was followed up for 7 months. She reported remarkable decrease in hirsutism and some improvement in voice. Laboratory levels of fasting insulin decreased to 47.3 mU/L.

**DISCUSSION**

This 13½-year-old girl had not attained menarche but has had pubarche and thelarche 1 year back, so she cannot be diagnosed as a case of primary amenorrhea but her symptoms and signs in the form of clitoromegaly and hirsutism warrant urgent evaluation.

Considering history, examination, and investigations, a provisional diagnosis of early-onset PCOS was made but dilemma arose, speculating severity of hirsutism, clitoromegaly, and raised levels of 17-hydroxyprogesterone poststimulation. Also, presence of insulin resistance falls in favor of PCOS but normal levels of LH and FSH falls against it. On enquiring about presence of ambiguous genitalia at birth, her parents reported it to be normal. Thus, taking into consideration all the above facts, final diagnosis of NCCAH was made.

Even though both these entities require to be treated with antiandrogens, it is important to differentiate them from each other as NCCAH also requires additional glucocorticoid supplementation, although some cases of PCOS might require steroids. Another reason to diagnose it correctly is, during their reproductive period if her partner is also a carrier of CAH, the newborn may succumb to classical CAH.

Polycystic ovarian syndrome in the background of androgen overproduction (or persistent disturbances of the hypothalamic-pituitary-adrenal axis even after normalization of androgen levels) suggests adrenal enzyme defects, but the precise interaction of the ovary and adrenal cortex via serum steroids remains hazy. 4

Counseling of the patient and her parents is critical as due to androgen excess many women struggle with the loss of feminine identity and later infertility subsequently affecting their social life.

**CONCLUSION**

Thus, there is significant overlap between PCOS and NCCAH, which warrants accurate diagnosis based on hormonal analysis to institute early and appropriate therapy.
CLINICAL SIGNIFICANCE

Early therapy can prevent infertility and androgenic complications later in life.

REFERENCES


