Case reports

Untreated congenital adrenal hyperplasia presenting with severe androgenic alopecia

J B O'Driscoll MRCP[1] **D C Anderson** MD FRCP[2]
[1]*Department of Dermatology, The Skin Hospital, Salford,* [2]*Professor of Medicine, Prince of Wales Hospital, Hong Kong*

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A case of congenital adrenal hyperplasia presenting for the first time at the age of 59 with severe androgenic alopecia and marked virilization is reported.

Case report

This lady presented in October 1991 with a 30 year history of alopecia. She had previously attended a private 'hair clinic' where topical minoxidil had been prescribed but no diagnosis made.

She had always been 'hairy' as a child and up to the age of 8 years was the tallest girl in her class. She then stopped growing at her present height of 4 ft 11 in. She has never had periods nor have her breasts ever developed. From childhood she had been aware that she had 'been born with a sexual abnormality that nothing could be done about' and despite repeated contact with the medical profession for various unrelated conditions the problem had been ignored. She has been teased about her appearance throughout her life and tried to commit suicide at the age of 11 by drinking carbolic acid.

She started to go bald in her mid-twenties and this has been a major source of embarrassment although she has only worn a wig for the last year. She shaves her face twice a day. She has never had any problem with sexual identity but has never had a sexual relationship and has always been embarrassed that she has been propositioned by women at work. She has always remained in employment albeit working mainly on night shift.

On examination she was of short stature and very muscular. She was almost completely bald with a Hamilton grade V pattern (Figure 1). Her face was shaven and she was generally severely hirsute (Ferriman Gallwey score 39). There was a male escutcheon and a penis sized phallus with small vaginal orifice.

Serum testosterone was 25 nmol/l (normal range up to 2.4) with androstenedione 30 nmol/l (2.0-8.0) and baseline 17-hydroxyprogesterone (17-OHP) 520 nmol/l (<0.3-20). Plasma ACTH was 883 ng/l (<10-50). 21-hydroxylase deficiency was confirmed by urinary steroid profiling which revealed very high levels of the 17-OHP metabolites in particular pregnanetriol 15.4 mg/24 h (<1.5).

Computerized tomography scan showed both adrenal glands to be significantly hypertrophied. A high resolution ultrasound scan of the pelvis showed small but otherwise normal uterus and ovaries.

Comment

The clinical features are those of very severe virilization due to congenital adrenal hyperplasia (CAH)[1]. The grossly raised levels of 17-OHP confirm that this is due to 21-hydroxylase deficiency, an enzyme deficiency which accounts for 90% of cases of CAH[2,3]. Defects in the adrenal 21-hydroxylase gene impair the biosynthesis of both cortisol and aldosterone and lead to an accumulation of 17-OHP which is diverted into the pathways of androgen production thus leading to virilization[2,4]. She falls into the category of 'simple virilizing disease' and does not have the 'salt-wasting' form of the disease, in which aldosterone synthesis is insufficient to prevent hyponatraemia, from which two-thirds of patients with CAH suffer[2]. It is evident that abnormalities were present from the time of her birth and it is extraordinary for a patient to present undiagnosed at the age of 59 with such severe virilization. She is an intelligent woman and despite the great psychological problems caused by her condition she evidently decided that there was nothing that could be done about it and adapted as well as she could.

Treatment has been started with prednisolone 2.5 mg/mane and 5 mg nocte and serum androgen levels have been reduced to well within normal limits.

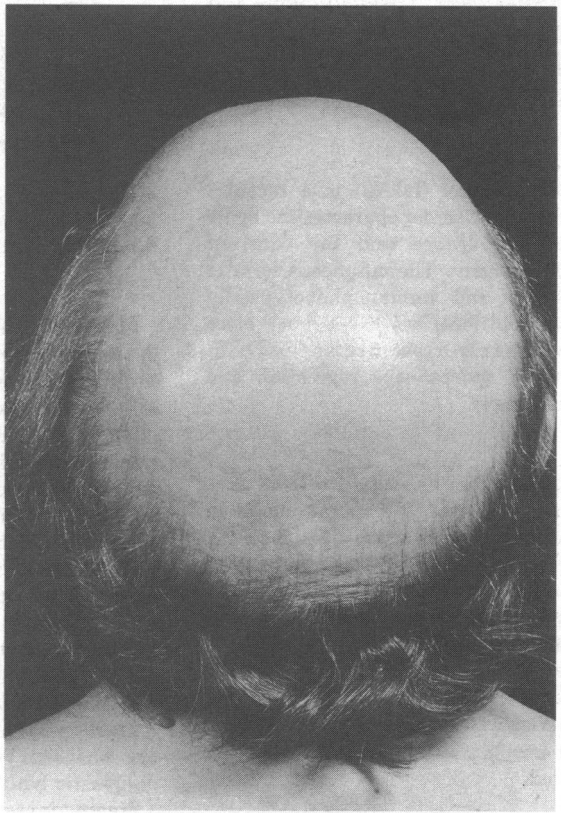

Figure 1. Marked male pattern baldness

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Correspondence to: Dr J B O'Driscoll, The Skin Hospital, Salford Royal Hospital, Chapel St, Salford M60 9EP

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