Atypical circumstances of 46,XY female diagnosis (case report)

Z. Haouchine*[a] (Dr), A. Vujovic[a] (Dr), J. Desir[a] (Pr), B. Corvilain[a] (Pr), N. Driessens[a] (Pr)
[a] Erasme Hospital, Faculty of Medicine, Université Libre de Bruxelles, Belgium, Bruxelles, BELGIQUE

INTRODUCTION

Complete Androgen Insensitivity Syndrome (CAIS) is a rare cause of primary amenorrhea with normal breast development.
It is usually diagnosed at the adolescence after consultation due to primary amenorrhea

CASE REPORT

A 35-year-old woman consulted a dermatologist for hair loss, without mentioning any sexual disorder.
She was referred to the endocrinologist after measurement of an increased serum total testosterone above 8-fold the upper value of the normal range [6.5 ng/ml (N: 0.2-0.8)].
Anamnesis reported a primary amenorrhea and a consanguineous family.
At clinical examination, she was slightly overweight (height: 167 cm; weight: 77 kg; BMI: 28 kg/m^2) with a normal breast development and adult external genitalia but no axillary or pubic hair. There was no hirsutism nor acne nor hyperseborrhea.
The patient has a female phenotype without ambiguity and a female sexual and social identity.

Biology showed normal 17-hydroxy-progesterone (1.12 ng/ml) and elevated AMH [98 ng/ml (N: 1.5-6.5)].

Pelvic ultrasonography revealed an absence of uterus, a short vagina and bilateral intraabdominal gonads without follicles nor individualized tumor.

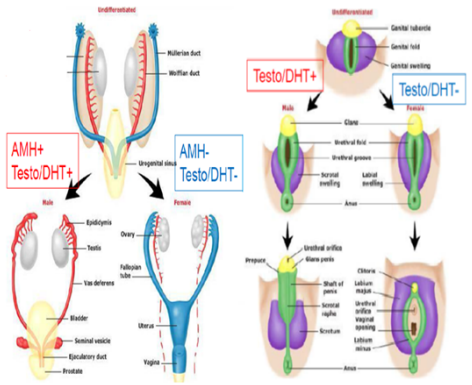
DISCUSSION

The absence of sexual hair contrasted with the high levels of total testosterone suggesting an androgen resistance. The elevated AMH suggested a testicular origin and explained the absence of uterus.(1)

Caryotype of 46XY confirmed the suspected diagnosis of complete insensitivity to androgens syndrome (CAIS).
The genetic analysis demonstrated a hemizygous carrying for the c, 598G>T, p,Glu200Ter mutation in exon 1 of the Androgen Receptor gene.
This introduces a stop codon resulting in a non functional protein.

PHYSIOPATHOLOGY :

In the absence of androgenisation (no testosterone or receptor deficiency), the external genitals are female (2,3)
The absence of uterus is due to AMH produced by the functional testis. It induces the involution of the Mullerian ducts. (2,3)



CONCLUSION

The particularity of the present case is the atypical circumstances of diagnosis which demonstrates the importance of the psychological care and the need of a multidisciplinary team.
The medical management consists of bilateral gonadectomy after pubertal induction in order to prevent the risk of gonadal cancerisation (4). It has been proposed to the patient but it has not been done yet.

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