COMPLETE ANDROGEN INSENSITIVITY SYNDROME IN A 21-YEAR OLD, A CASE REPORT

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INTRODUCTION

Complete androgen insensitivity syndrome (CAIS) is a rare X-link disorder caused by mutation in androgen receptor gene on chromosome Xq11-12 (1). Due to this resistance to androgens people with CAIS have 46XY karotype, however are phenotypically female with no uterus and undescended testes (2). Initial presentation is commonly seen during infancy with inguinal masses or hernias containing testes, or in post pubertal women with primary amenorrhea (3). Management commonly involves hormonal replacement and gonadectomy postpubertally to reduce risk of developing germ cell tumours.

CASE

This case report describes a 21-year-old phenotypical female who presented to the emergency department with post coital bleeding requiring examination under anaesthesia. On subsequent examination a blind vagina pouch of 4cm with a small posterior wall laceration was observed, with no palpable uterus on bimanual examination. Ultrasound demonstrated the absence of uterus or ovaries which was further confirmed of MRI with the presence of undescended testes. Karyotyping and FISH analysis detected male karyotype 46XY, with no evidence of mosaic sex chromosome aneuploidy.

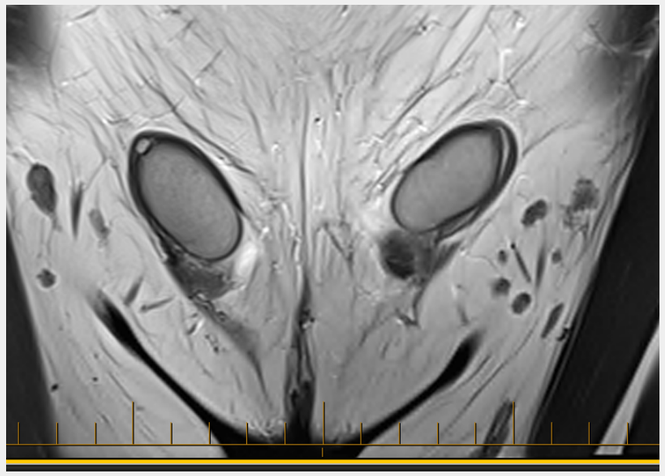

Figure 1: MR Pelvis of patient with CAIS, demonstrating undescended testes

OBJECTIVE

The aim of this case report is to highlight the incidental late diagnosis of CAIS in a 21-year-old who presented with vaginal bleeding.

MANAGEMENT

The patient opted for bilateral open orchidectomy to reduce her risk of malignancy which was facilitated by the General Surgeons. She was followed as an outpatient post operatively and commenced on topical estrogen hormonal replacement. She was referred for psychological support, and physiotherapy where she was prescribed vaginal dilators to help increase the length of her vagina.

DISCUSSION

The diagnosis of CAIS is usually seen at puberty with primary amenorrhea, however in this case diagnosis was delayed due to patient embarrassment in seeking guidance regarding the absence of regular menses. Long term medical and psychological management is important as sexual dissatisfaction is commonly reported and for monitoring of bone mineral density.

References:

1. Lubahn DB, Joseph DR, Sullivan PM, Willard HF, French FS, Wilson EM. Cloning of human androgen receptor complementary DNA and localization to the X chromosome. Science. 1988 Apr 15;240(4850):327-30. doi: 10.1126/science.3353727. PMID: 3353727.
2. Doehnert, U., Bertelloni, S., Werner, R., Dati, E., & Hiort, O. (2015). Characteristic features of reproductive hormone profiles in late adolescent and adult females with complete androgen insensitivity syndrome. Sexual Development, 9(2), 69–74. https://doi.org/10.1159/000371464
3. Tyutyusheva, N., Mancini, I., Baroncelli, G. I., D'Elios, S., Peroni, D., Meriggiola, M. C., & Bertelloni, S. (2021). Complete androgen insensitivity syndrome: From bench to bed. International Journal of Molecular Sciences, 22(3), 1264. https://doi.org/10.3390/ijms22031264