

Unusual Presentation of Sertoli Cell Only Syndrome with Tiredness

Authors: Dr M Razeem, Medical Registrar; Dr Z Choudhry, CTM2; Dr F Haddadin, Consultant Endocrinologist

Queen Elizabeth Hospital King's Lynn

OBJECTIVES

Sertoli Cell Only Syndrome (SCOS) is a rare cause of male infertility in which the cause is often unknown. There has been poor reporting of symptoms in SCOS except commonest investigating cause of infertility. We hope sharing this case will help broaden our understanding of atypical presenting features of SCOS.

CASE REPORT

Here, we report an unusual presentation of SCOS in a 32 year old man with severe fatigue, flushing, aches and hypothyroidism. These symptoms did not improve despite correcting hypothyroidism with thyroxine replacement. There have been no features of depression and he has been investigated thoroughly as predominant fatigue has been affecting everyday life. Patient is married and has no children. Bedside observations and general physical examination is normal. Laboratory testing for anaemia, kidney function, Vitamin D, liver function, coeliac screen, vasculitic screen and anti-acetylcholine-receptor-antibody is negative. Plain X-ray of chest is normal. Pituitary axis testing revealed raised Follicle Stimulating Hormone (FSH) of 34.4 IU/L (normal < 8 IU/L), borderline Lieutininising Hormone (LH) of 8.5 IU/L (normal 3-8 IU/L), normal prolactin, normal levels of morning cortisol and normal Short Synacthen Test. His semen analysis confirmed Azoospermia. Patient is diagnosed with probable SCOS and is being considered for biopsy.

TOPIC

SCOS often presents as men (usually 20-40 years) seeking medical attention for infertility and are often suspected with history of infertility and azoospermia on semen analysis. However there are reported cases of presentation of SCOS with ambiguous genitalia at as young as at childhood¹. Patients usually present with small to normal testes and normal physical appearance (without gynacomastia), however there are few reported cases with testicular ectopia (Ozdemir et al). The causes of SCOS could be any cause of spermatogenic failure, iatrogenic causes such as toxins and radiations and also genetic and hormonal factors. Sex chromosomes are involved in more than 23% of patients³. In most cases, a specific cause is not often found or persued due to poor prognostic benefit. Patients usually have normal plasma testosterone levels and therefore most useful laboratory test is the plasma FSH level as an elevated FSH level of >2 times upper limit of normal indicate spermatogenic failure and is an indication for biopsy. Imaging studies is not useful in diagnosis of SCOS and therefore testis biopsy is the investigation of choice for definitive diagnosis. Clinically it is important to be mindful that for patients with high suspicion of clinical and laboratory diagnosis of likely SCOS (if FSH>2-3 times normal range), a biopsy is not helpful in management if the patient does not wish to be considered for Intra-Cytoplasmic Sperm Injection. Though one could argue that once a diagnosis is made, imagings along with biopsy findings are useful for screening for at risk conditions such as testicular malignancy. There is no effective medical treatment for SCOS and surgical management for fertility such as Test will only benefit a small subset of patients with SCOS who may have an isolated foci of spermatogenesis within a testes.

DISCUSSION

A testis biopsy is not indicated in this case for the management of SCOS itself as the patient does not wish to have any children. However it could be useful for risk assessment for testicular cancer and should be guided by patient's wishes. Although there is no medical treatment for SCOS, a trial of treatment based on presenting symptoms and laboratory findings could be commenced to see if there is any life style or symptomatic benefit. The reported patient had disabling tiredness resulting in loss of job and relationships and therefore anything to help with his tiredness will probably benefit him most. There is rapid advancement in the genetic side of SCOS and genetic causes are common³. Therefore it is important to perform genetic testing and karyotyping for patients considered to be assisted fertilisation as this will allow appropriate prior genetic counselling if an inherited cause is to be found.

REFERENCES

1. Gurbuz, F et al. Sertoli cell only syndrome with ambiguous genitalia. *Journal of Pediatric Endocrinology & Metabolism*; Berlin Vol. 29, Iss.7, (2016): 849-852.
2. Özdemir E; Koçan H; Yazıcı M; Altıok E; Su FE et al. Case Report of a Man With Sertoli Cell Only, Transverse Testicular Ectopia, and External Auditory Canal Atresia. *Urology case reports*; Jan 2015; vol. 3 (no. 1); p. 3
3. Stouffs K et al. Sertoli Cell-Only Syndrome: Behind the Genetic Scenes. *Biomed Res Int.* 2016; 2016: 6191307.