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Case Report

Sertoli Cell-Only Syndrome with Normal Karyotype

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Abstract

Sertoli cell-only syndrome is usually associated with micro deletion Y chromosome. We reported rare case in the 37years-old-man married 10 years ago. A left varicocele was associated with this syndrome. No micro deletion Y chromosome was noted. Left varicocelectomy has been perfomed.

Keywords: Azoospermia; Sertoli Cell-Only Syndrome; Testicular Biopsy.

Introduction

The seminiferous epithelium contains both somatic cells (Sertoli cells) and germ cells (from the early stem cells to spermatozoa). Sertoli cells perform crucial functions that initiate and sustain spermatogenesis. Consequently, an abnormality in their function may disrupt the full progression of spermatogenesis [1,2]. Azoospermia due to spermatogenic failure, or non-obstructive azoospermia, affects approximately 1% of the general population and 10%-15% of men seeking an infertility evaluation [3,4].

Case Report

A 37 year-old-man patient presented with primary infertility. He was married 10 years ago. He was not decreased libido or erectile dyfunction. His medical (chlamydia, Mycoplasma, syphilis, tuberculosis) and surgical history were negative. On examination, height was 175 cm, weight 74 kg. Body hair was normal. Sexual maturing rating was normal with a testicular volume bilaterally. He had not gynecomastia. Biological screening shows creatinine level was 6 mg/L, hemoglobin was 14 g/dL, and white blood cell count was 6 G/L. Semen analysis showed azoospermia on three consecutive samples even after centrifugation. Doppler color ultrasound of the scrotum showed left varicocele. The testicular, the epididymis and the vas deferens was normal.

Serum Follicle-Stimulating Hormone (FSH) level was 45,93 mIU/ml [1-5], Serum Luteinizing Hormone (LH) level -13,14 mIU/ml [1-8] and total serum testosterone - 4,26 ng/ml [2,5-8]. Serum Prolactin level - 15,4 ng/ml [3-25]. Patient has a normal Karyotype (Normal 46, XY karyotype, as shown Giemsa-trypsin [G-banding]. Left varicocelectomy has been perfomed and testicular biopsy. The postoperative course was uneventful. His testicular biopsy revealed Sertoli Cell-Only Syndrome (SCOS). However, the patient done an Intracytoplasmic Sperm Injection (ICSI) after Testicular Sperm Extraction (TESE) successfully to be father.

Discussion

Primary testicular failure occurs in approximately 1% of all males, and is present in 10% of those obtaining medical consultation for infertility [5]. Sertoli cell-only syndrome (SCOS), [5,6] also known as germinal cell aplasia, can be found in a few testicular biopsies that are performed in these patients. The pathognomonic feature of SCOS is the absence of germ cells. The seminiferous tubules are lined by Sertoli cells that may be either immature (prepubertal) or mature or have other changes that may correlate with specific etiologies and clinical findings [7]. For the prognostic purpose, Anniballo et al. [5] divided SCOS into two categories: Pure (congenital) and mixed (secondary). The pure form is caused by failure of migration of germ cells, [8] while the mixed form is related to the postnatal damage to previously healthy testicular tissue. Retrieving germ cells in cases of pure SCOS is the impossible [9] and proper identification of the type of SCOS is essential to spare unnecessary medical expense and inconvenience to patients. The combination of increased inhibin B and normal serum FSH levels also indicates the presence of spermatids [10]. However, the role of inhibin B in predicting the presence of sperm within the testis in patients with NOA is still controversial [11].

Although some cases of azoospermia due to SCOS (approximately 10%-15%) are due to microdeletions of variable portions of the AZF region of the Y chromosome, the etiology of most is unknown. The correct identification of infertile patients with true SCOS by using selective molecular markers for germ cells might help in establishing the true incidence of this problem and in directing research toward understanding the causes of this defect [12]. Many investigators have suggested that as more genes are deleted, spermatogenesis defects may progress from hypospermatogenesis to SCOS. Cases with extensive deletions involving AZFa, AZFb, and AZFc are almost invariably associated with SCOS [13]. Ying Hui Lin et al. said [14], this case had deletions of the whole long arm of the Y chromosome, and only Sertoli cells were found in the testicular specimen, a finding in concordance with previous reports [15,16].

However, many of these patients have a normal XY karyotype with normal secondary male sexual characteristics, yet are infertile and azoospermic or severe oligozoospermic. karyotype. (Normal 46, XY karyotype, as shown Giemsa-trypsin [G-banding], but on polymerase chain reaction amplification or fluorescence in situ hybridisation microdeletions of the Y chromosome are seen, rarely macroscopic deletions of the Y-chromosome long arm that are detectable by karyotyping [i.e. 46, X, Yq-]) [7]. Predictive factors for the presence of spermatozoa within the testis have been actively studied, including serum FSH and inhibin-B levels, testicular volume, and testicular histopathology [17-18].

The analysed patients with Sertoli cell only syndrome have significantly elevated serum FSH level. The distincly elevated basal serum LH levels and serum testosterone levels in normal range, demonstrated that there was compensated disfunction of the Leydig cells in patients with Sertoli cell only syndrome. The elevation of gonadotropins in patients with this condition was not so high and levels of serum testosterone not lowered, as those seen in the patients with Klinefelter's syndrome [19] Suominem et al. [20] conclued that there is not any relation between testicular morphology (i.e. spermatogenesis) and serum prolactin, that increased prolactin values are usually associated with low testosterone values and that not correlation exists between serum gonadotrophin and prolactin levels. Intracytoplasmic Sperm Injection (ICSI) and with testicular sperm extraction (TESE) can be useful in retrieving sperm from these men with mixed SCOS [21,22].

Conclusion

The sertoli cell-only syndrome is often of genetic origin with micro deletion of the Y chromosome. It's often associated by the serum FSH, LH and serum testosterone level rising. This micro deletion may not exist but exceptionally.

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