Case Report

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Robertsonian Translocation Associated with Azoospermia

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Key Words

Robertsonian translocation

Infertility, male

Azoospermia

Abstract

We report a case of Robertsonian translocation associated with azoospermia. The literature is reviewed, and testicular histology and hormone condition are discussed.

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Introduction

It is well-known that sex chromosome abnormalities are related to male infertility [1]. Recently, a possible relationship between autosomal translocation and male infertility is emphasized [2, 3]. However, literature dealing with coincidence of azoospermia with Robertsonian (central fusion) translocation is scanty [4]. Herein we report a case of Robertsonian translocation associated with azoospermia.

(13ql4q) (fig. 1). Bilateral testicular biopsy was performed for the evaluation of spermatogenesis. Tubular walls were slightly thickened. Spermatogenesis was generally arrested at the stage of primary spermatocyte (fig. 2). In some seminiferous tubules, germ cells were disorganized and were detached from the epithelium (fig. 3). Neither spermatids nor spermatozoa were observed. However, Leydig and Sertoli cells appeared normal.

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Case Report

A 30-year-old Japanese man was referred to our hospital because of his infertility. He was a well-developed male weighing 63 kg with a height of 165 cm. Physical examination revealed male habitus with normal adult pubic and axillary hair. Neither malformations nor gynecomastia were seen. The penis, epididymides, spermatic cords and prostate were normal. Both testes were of normal size and consistency. The right testis was about 22 ml in volume and left one was approximately 19 ml with respective orchidometer measurements. Repeated spermatograms revealed the absence of spermatozoa. Laboratory investigations including blood analysis,

urinalysis and blood chemistry were within normal ranges. Plasma luteinizing hormone was 20 mIU/ml (normal range, 5–30 mIU/ml), follicle-stimulating hormone 8 mIU/ml (normal range, 5–22 mIU/ml), testosterone 4.8 ng/ml (normal range, 3.0–8.5 ng/ml) and prolactin 11 ng/ml (normal range, 2–20 ng/ml). Chromosome analysis of peripheral lymphocytes revealed a karyotype of 45, XY, -13, -14, +t

Fig. 1. Chromosome analysis reveals a karyotype of 45, XY, -13, -14,+t(13q14q).

380

Sasagawa/Nakada/Terada/Katayama

Fig. 2. Seminiferous epithelium shows maturation arrest in spermatogenesis. HE. $\times 150$.

Fig. 3. Germ cells are disorganized and are detached from seminiferous epithelium. HE. $\times 150$.

Discussion

Robertsonian translocation associated with azoospermia is a rare condition. Buurrouillou et al. [5] reported 1 case of Robertsonian translocation among the 383 azoospermic men. In our department only 1 case has been observed among the 119 patients with azoospermia.

Several theories concerning the mechanism of autosomal translocation on spermatogenic failure have been proposed; position effect [6], low chiasma count [7], impaired synapsis [8] and perturbed inactivation of the YX bivalent [9]. However, formal evidence for these theories is still lacking.

Histological studies on testicular biopsies from patients with Robertsonian translocation show partial or complete arrest of spermatogenesis and slight hyperplasia of Leydig cells [4, 10]. In our case, spermatogenesis

was arrested at the stage of primary spermatocyte, while Leydig and Sertoli cells appeared normal. Plasma hormone levels were within normal ranges in our case. In patients with sex chromosome abnormality, severe hyalinization of seminiferous tubules and hyperplasia of Leydig cells are usually observed [11]. Furthermore, hyper-gonadotropic hypogonadism is common in sex chromosome abnormality [12]. These facts suggest that histological changes of the testis and hormonal abnormalities are milder in Robertsonian translocation than in sex chromosome abnormality.

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