Case Report

Received: June 26, 1989

Accepted: July 6, 1989

Urol Int 1990;45:181-182

A Case of Klinefelter's Syndrome with Bilateral Absence of the Vas Deferens

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Key Words

Klinefelter's syndrome

Vas deferens, absence

Abstract

A 33-year-old male presented to the Chiba University Hospital with the main complaint of right flank pain. Bilateral vas deferens were not palpable. Hormonal examination revealed hypergonadotropic hypogonadism and cytogenetic studies a 47, XXY karyotype. The significance of the association of this karyotype with the absence of vas deferens is discussed.

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Introduction

Abnormality of the external genitalia or internal genital duct is rare in Klinefelter's syndrome. Leiba et al. [1] first reported a case of Klinefelter's syndrome with bilateral absence of the vas deferens. Thereafter no other case of Klinefelter's syndrome with this anomaly has been reported. Herein, a case with these two malconditions is presented.

smaller increase to 110.1 mIU/ml at 30 min after an injection of 100 µg luteinizing hormone-releasing hormone.

Lymphocyte cultures followed by G and Q banding showed a chromosome complement of 47, XXY (fig. 1). Semen analysis showed azoospermia. Intravenous pyelography revealed normal findings.

From the above findings, the diagnosis of Klinefelter's syndrome with bilateral absence of the vas deferens was made and testicular biopsy and examination of vas deferens were performed. Bilateral normal epididymis were found but the vas deferens was absent in both sides. Histology of both testes obtained by biopsy revealed hyalinization of the seminiferous tubules and proliferation of Leydig cells (fig. 2).

Case Report

A 33-year-old male presented to the Chiba University Hospital with the main complaint of right flank pain. At the patient's birth, his father and mother were 30 and 27 years old, respectively. Pregnancy and delivery were normal and there was no disease in his past history. He was 167 cm tall and 67 kg in weight. His arm span was 173.5 cm. He had pubic hair of male type. The

volumes of the testes was 3 ml each. Bilateral vas deferens were not palpable. Rectal examination showed a small sized prostate gland. There was no gynecomastia. Hematological and liver function tests and urinalysis were within normal range. The basal levels of luteinizing hormone (LH), follicle stimulating hormone (FSH) and testosterone in blood were 56.0 mIU/ml, 49.7 mIU/ml and 285 ng/dl, respectively, which correspond to hypergonadotropic hypogonadism.

After administration of human chorionic gonadotropin (hCG, 4,000 IU) for 3 days, the testosterone level in blood rose from 285 to 300 ng/dl. LH rose to 290.0 mIU/ml at 30 min and FSH showed a

Discussion

It has been reported that of all males investigated for infertility, 0.3–1.5% have no vas deferens [2–5] and 5–10% of the men with azoospermia have this anomaly [6, 7], which means that it is not a rare cause of male infertility.

Michelson [3] postulated that absence of the vas deferens must be the result of a disturbance of the outgrowth of the Wolffian duct, which is controlled by testosterone secreted from embryonic testes. Then hormonal disorders in gestation may cause this anomaly, but abnormality of internal genital duct in Klinefelter's syndrome is very rare [1], which revealed most of the

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Fig. 1. Chromosome analysis with peripheral blood revealed 47.XXY.

Fig. 2. Histology of both testes showed hyalinization of seminiferous tubules and proliferation of Leydig cells.

cases of Klinefelter's syndrome had sufficient function of Leydig cell for the normal development of the internal genital duct. In the present case, the blood level of testosterone is slightly decreased, which showed hormonal disorders were less attributable to this anomaly. This hypothesis would explain the absence of vas deferens, epididymis and seminal vesicle. However, in the case of total or partly epididymis, this anomaly may be the result of atrophy of a previously normally developed vas deferens. The patient with fibrous strands in place of the vas deferens seems to support this possibility [8]. According to the literature [9, 10], unilateral agenesis of the vas deferens is often associated with renal malformation, usually aplasia of the kidney on the affected side. Therefore, intravenous pyelography should be performed. Vasovasostomy, epididymovasostomy, creation of artificial spermatocele and so on are sometimes performed to obtain fertility [11]. But in the present case, it was no use to try such procedures, since there was no spermatogenesis in both testes.

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