Case Report

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Klinefelter's Syndrome with Hypospadias and Bilateral Cryptorchidism

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Key Words

Klinefelter's syndrome

Hypospadias

Cryptorchidism

Abstract

A 3-year-old boy was found to have abnormality of the external genitalia at birth. Physical examination revealed hypospadias penis and bilateral cryptorchidism. Chromosomal analysis of peripheral blood showed the karyotype of 47, XXY, and the diagnosis of Klinefelter's syndrome associated with hypospadias and cryptorchidism was made. Klinefelter's syndrome is rare in infancy.

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Case Report

The patient, a 3-year-old boy, was found to have ambiguous genitalia at birth. He had been born in November 26, 1982, with hypospadias penoscrotalis and bilateral cryptorchidism. Mother and father were 21 and 25 years old when he was born. His birth weight has been 2,770 g after a 41-week gestation. The mother had not taken any drugs during pregnancy. Chromosomal analysis of the peripheral blood showed the karyotype with 47, XXY. He had been discharged, but would receive one-stage urethroplasty and orchio-pxy in the future.

In 1985, the boy was admitted to the Yokosuka Kyosai Hospital for evaluation and receiving therapeutic procedures. He had hypospadias penis and bilateral cryptorchidism on physical examination. Mental retardation was not detected. Excretory urograms showed a normal upper urinary tract. Serum luteinizing hormone, follicle-stimulating hormone, and testosterone levels were 11.6 and 2.8 IU/l and 26 ng/dl, respectively, and were within normal limits. Chromosomal analysis was performed again. Karyotyping showed 47, XXY. Mosaicism was not found. Other laboratory parameters were normal. The diagnosis of Klinefelter's syndrome with hypospadias and bilateral cryptorchidism was made. One-stage operation, Dacket's modified flip-flap procedure, to repair hypospadias was performed on November 29, 1985. The indwelling catheter was removed on the 10th postoperative day. Urinary leakage was seen at anastomosis of old and new urethra, but disappeared about 4 months later. He will receive bilateral orchiopey with testicular biopsy in the near future.

Discussion

Klinefelter et al. [1] in 1942 described a group of male patients with small testes, azoospermia, and gynecomasia. The clinical features of Klinefelter's syndrome are abnormality of external

genitalia, tall height, gynecomastia, and elevated gonadotropin levels [2]. Today, the definition of Klinefelter's syndrome is as follows: congenital anomaly with somatic cells with more than 2 X chromosomes and more than 1 Y chromosome [3]. The ratio in the newborn is approximately 1:500 [4], but it is rare with hypospadias or cryptorchidism [5], rare in infancy [6]. Previously, only 2 cases of Klinefelter's syndrome with hypospadias and cryptorchidism in infancy were reported [7, 8].

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