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46XX male; report of case

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RIGHT:
46XX MALE; REPORT OF CASE

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Chromosomal studies on a prepubertal 7-year-old boy with a normal male phenotype except for undescended testes showed a 46XX normal female karyotype. An H-Y antigen assay done at the same time on peripheral blood was positive in a titer similar to that of normal male subjects. Although the microscopic findings for both testes were normal, hypergonadotropic hypogonadism was suggested from both LH-RH and hCG stimulation tests.

Key words: 46XX male, H-Y antigen, Undescended testis

INTRODUCTION

In 1964, De La Chapelle and his group described the first case of the 46XX male syndrome\(^2\). Since then, an additional 135 cases have been reported in the world literature and 18 cases have also been recognized in the Japanese\(^3,4\). The clinical findings of the 46XX male syndrome consist of sexual ambiguity in the neonate and hypogonadism and infertility in the adult. We describe the 19th Japanese case of a prepubertal 46XX male with bilateral undescended testes.

CASE REPORT

A 7-year-old male patient was referred to our hospital for surgical treatment of bilateral undescended testes. He had been born at full term and his weight at birth was 2,700 g. The parents were not related, and the mother was 34 and the father 33 years of age at the time of his birth. The mother neither took drugs or was exposed to radiation during pregnancy.

On physical examination, he was a normal boy in phenotype except for defects of both scrotal contents. His height was 117.6 cm and he weighed 19.4 kg, which
Fig. 2. Serum basal and peak, LH, FSH and testosterone levels in LH-RH and hCG tests. Blood samplings were performed within 120 min. after LH-RH administration (LH-RH 100 μg/kg, i.v.) and 4 days after hCG administration (hCG 10,000 IU/m², i.m.). Shaded areas indicate mean ± 2SD in normal prepubertal boy.

Fig. 3. Karyotype from peripheral lymphocyte shows 46XX pattern.

are within the normal Japanese male ranges. The bone age correlated with the chronological age and neither cardiovascular nor skeletal anomalies could be detected. He had a normal dermatogram pattern and mental development was normal (Fig. 1). The excretory urogram was normal, and no vaginal remnant was found on a retrograde urethrocytogram. The results of various laboratory examinations, including a hemogram, blood chemistry, urinalysis and urinary excretion of 17-ketosteroids, were within the normal limits. Hypergonadotropic hypogonadism was suggested
from the serum levels of gonadotropins and testosterone after administration of LH-RH and hCG (Fig. 2).

The X body pattern from buccal smears was positive (29%). Chromosomal studies, which were performed on two separate occasions from cultured peripheral lymphocytes and on one occasion from scrotal skin, revealed a normal 46 XX chromosomal complement (Fig. 3). H-Y antigen assay from the peripheral blood was positive in a titer similar to that for normal male subjects (Fig. 4).

Exploratory laparotomy revealed no Müllerian structures. The right testis (1.2 × 1.0 × 1.0 cm in size) retained in the abdominal cavity was resected and the left testis (1.5 × 1.2 × 1.2 cm in size) retained in the inguinal canal was fixed in the scrotal pocket after biopsy. Microscopic examination of specimens from both testes revealed an immature testis showing seminiferous tubules without spermatogenesis (Fig. 5).

DISCUSSION

Testicular differentiation has been associated with the presence of the H-Y antigen, a cell surface component coded for by genes in the pericentric region of the Y chromosome. In fact, the testicle does not exist in the absence of serologically detectable H-Y antigen, which is the inducer of the mammalian testis. In our patient, despite the presence of a chromosomal constitution of 46XX, both testes and no Müllerian remnants were found. Thus, these testes had apparently been able to cause regression of the Müllerian ducts during fetal life.

The 46XX male syndrome is a rare entity in which hypogonadism is common, yet associated genitourinary malformations are rare. Undescended testis has been reported in 15 per cent of these cases. In our patient stimulation with a human chorionic gonadotropin resulted in a poor response in serum testosterone in contrast to the uniformly sharp response to pituitary gonadotropin previously reported. This finding may be due to the variable extent of testicular dysgenesis recognized in this syndrome as in 47XXY Klinefelter's syndrome. Because gonadal dysfunction in terms of andrology and spermatogenesis is supposed to become more obvious during puberty despite showing normal infantile testes in the prepubertal period, a long follow up is needed.

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和文抄録

46XX男性の一例

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両側精巣の他は正常男性型を示す7歳男児の染色体所見が60XX染色体を伴い、表皮にY染色体を示した。LH-RH および hCG 負荷試験にて高GH血症が示され、46XXの正常女性型であった。また末梢血中の H-Y 抗原も陽性を示し、Y染色体の性差栄養機能障害が示唆された。