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CASE REPORT

# Androgen Insensitivity Syndrome (Testicular Feminization)

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## Introduction

Androgen insensitivity (testicular feminization) syndrome is a rare inherited form of male pseudohermaphroditism that occurs in phenotypically normal women with adequate breast development, normal external genitalia, a vagina of variable depth, absent uterus, and sparse or absent pubic hair and axillary hair. These patients have male karyotype (XY) and negative sex chromatin. The gonad (undescended testis) may be intraabdominal, inguinal, or labial [1]. The incidence of testicular feminization syndrome is reported to range between one in 2,000 to one in 62,400 [2]. The present case is the first one from Jammu and Kashmir State of India where chromosome study has been carried out and 46, XY karyotype has been detected in the phenotypic female.

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## Case History

A 20-year-old phenotypic female clinically diagnosed as a case of primary amenorrhea was referred for chromosome study. There was no family history of any chromosomal abnormality in the family; hence, it was an isolated case. The patient was 169 cm tall and weighed 68 Kg. On physical examination she showed normal breast development but scanty pubic and axillary hair growth. Ultrasonographic study showed agenesis of uterus, however, ovaries could not be visualized. MRI of the pelvic region was performed to obtain the clear status of the uterus and ovaries. MRI report showed absence of uterus and ovaries but two well formed, normal sized testes were visualized. Left testis was seen in the labia majora whereas right testis was in the inguinal canal (Fig. 1a, b). Both the testes were removed surgically and they were submitted for histopathological study.

Preoperative endocrinological analysis showed extremely high level of Testosterone (837.09 ng/dl) whereas the level of other hormones was within normal range. Invasive surgery was planned and postoperative endocrinological analysis was advised only after karyotypic analysis was made. Postoperative endocrinological analysis showed the level of testosterone and other hormones to be within the normal range but the level of serum dehydroepiandrosterone, was high (12.36 ng/ml). After surgery, testes were submitted for histopathological study through histopathological slides. Atrophied seminiferous tubules along with marked Leydig hyperplasia were detected in both the testes

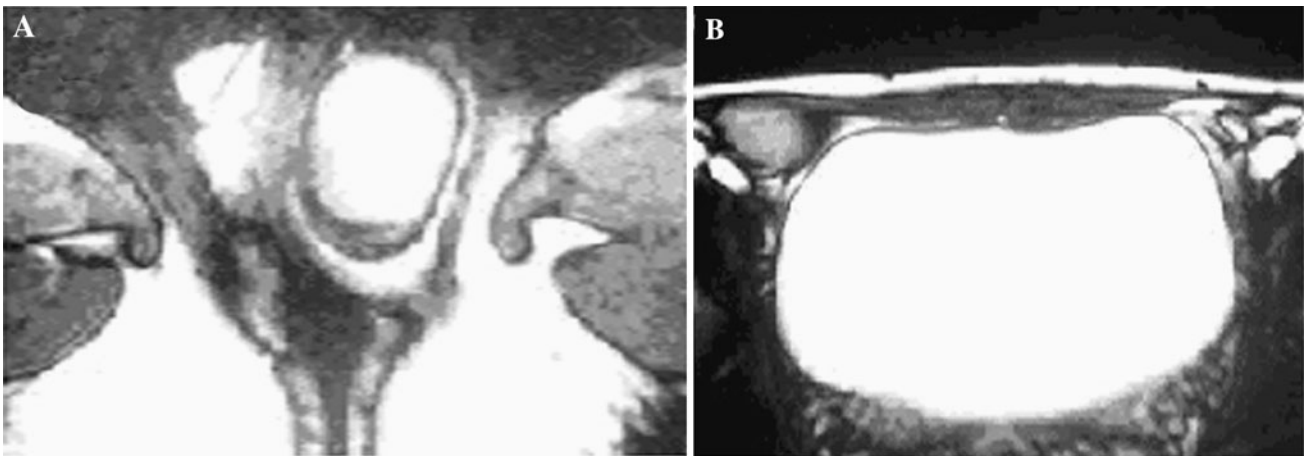


**Fig. 1** **a** MRI showing left testes in the labia majora. **b** MRI showing right testes in the inguinal canal

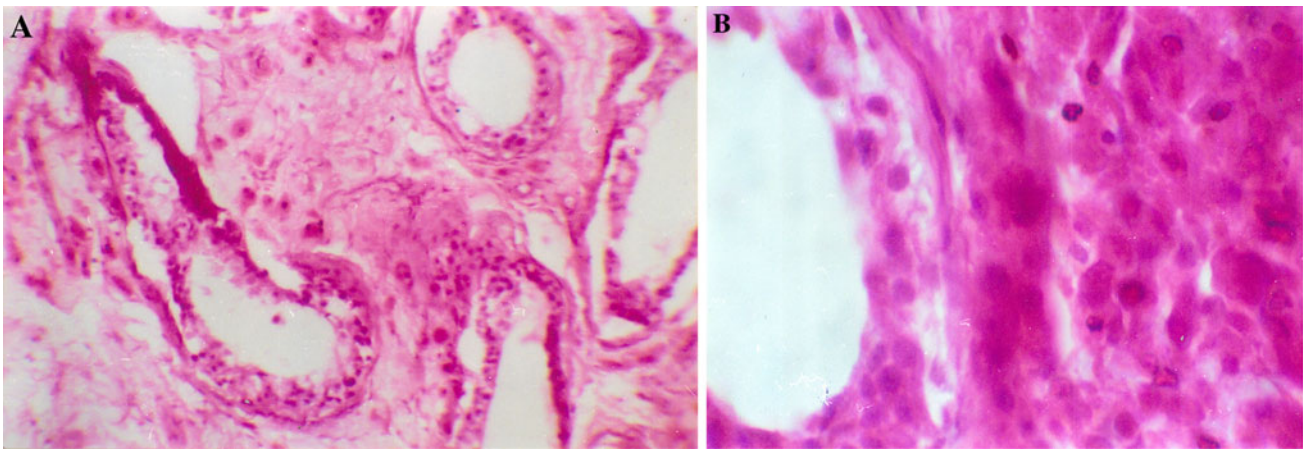


**Fig. 2** **a** Atrophied seminiferous tubules with thickened basement membrane (100×). **b** Leydig cell hyperplasia

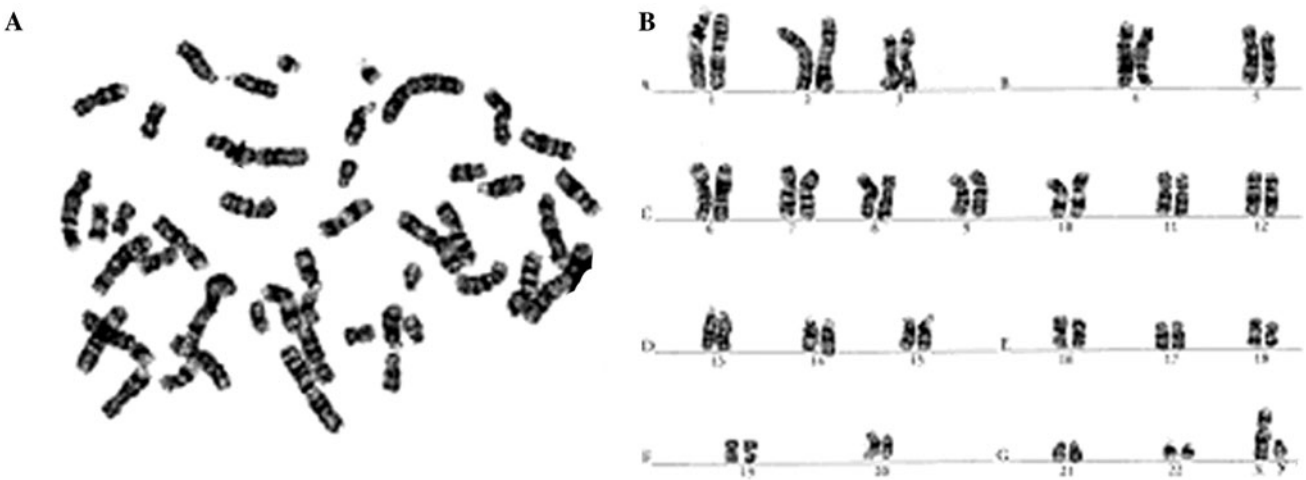


**Fig. 3** **a** and **b** Metaphase plate and karyotype of the subject

(Fig. 2a, b). Similar histopathological findings were obtained by Gad El-Moula et al. [3] in a female with androgen insensitivity syndrome.

Although the ultrasonography, MRI, and hormonal assay were in favor of the referred patient to be a case of testicular feminization, the final opinion was left to the chromosome study.

Slides prepared from lymphocyte culture were scanned. Some of the G-banded metaphase plates were selected for the preparation of their karyotypes. Every G-banded metaphase plate contained 46 chromosomes (Fig. 3a).

Karyotypes were prepared from the metaphase plates. Every karyotype contained 44 autosomes and a pair of sex chromosomes, 46, XY. Thus every karyotype had the sex-chromosome constitution of a male, 46, XY (Fig. 3b).

Therefore, through chromosome study and karyotypic analysis, the referred patient was confirmed to be a case of testicular feminization, a phenotypic female possessing the sex-chromosome constitution of a male (XY).

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