Case Report

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Androgen Insensitivity Syndrome (Testicular Feminization) – A Retrospective Diagnosis: Case Report

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ABSTRACT

Testicular feminization, or the androgen insensitivity syndrome, is a rare disease. It occurs due to various abnormalities of the X chromosome, a male, genetically XY, has some physical characteristics of a female or a full female phenotype. The androgen insensitivity syndrome occurs because of a resistance to the actions of the androgen hormones, which in turn switches the development towards the aspect of a female. We report a retrospectively diagnosed case of complete androgen insensitivity syndrome in a 50 years old woman who presented with right inguinal swelling. We aim to improve our knowledge of this illness from the data that provides us this study.

Keywords: *Testicular Feminization, Androgen Insensitivity, Retrospective.*

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. The incidence of testicular feminization syndrome is reported to range between one in 2,000 to one in 62,400. The present case is retrospectively diagnosed on the basis of incidental histopathological findings. Chromosome study has also been carried out and 46, XY karyotype has been detected in the phenotypic female.[1]

Case Report

A 50-year-old female presented with right inguinal swelling. Patient was clinically diagnosed as right inguinal hernia and was operated for the same. Intra operatively a retroperitoneal structure (?? nature) attached with hernial sac was found, same was excised and submitted for histopathology. Histopathological findings of the retroperitoneal tissue revealed testicular tissue with atrophied seminiferous tubules and thickened basement membrane. In view of histopathological findings, patient was called upon to perform physical examination and further investigations. As per the personal and past history – patient never had menstruation. Physical examination showed normal breast development but scanty pubic and axillary hair growth, the labia are small and hypoplastic, and the vulva is unpigmented. 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. Endocrinological analysis showed the high levels of testosterone (447 ng/dl). Luteinizing hormone (LH – 56.97 mIU/ml) and Follicular stimulating hormone (FSH – 64.03 mIU/ml) showed post-menopausal levels. The karyotype was mapped in order to differentiate the androgen insensitivity syndrome from other genetic abnormalities, like the Klinefelter syndrome (46XXY), Turner syndrome (45XO), mixed gonadal dyssynergia (45XO/46XY) or tetragametic chimerism (46XX/46XY). Karyotypic analysis of the patient confirmed the suspected diagnosis of testicular feminization, a phenotypic female possessing the sex-chromosome constitution of a male (XY).[2]

Discussion

Androgen insensitivity syndrome is typically characterized by evidence of feminization (i.e., under masculinization) of the external genitalia at birth, abnormal secondary sexual development at puberty, and infertility in individuals with a 46 XY karyotype. It represents a spectrum of defects in androgen action and can be subdivided into 3 broad phenotypes: (1) complete androgen insensitivity syndrome (CAIS) with typical female genitalia; (2) partial androgen insensitivity syndrome (PAIS) with predominantly female, predominantly male, or ambiguous genitalia; and (3) mild androgen insensitivity syndrome (MAIS) with typical male genitalia. The incidence of androgen insensitivity syndrome is estimated to be 1:20,000-64,000 male births.[3]

The first problem with Complete Androgen Insensitivity Syndrome is the diagnosis. It requires many tests, some quite rare and frequently unavailable, so therefore it is often

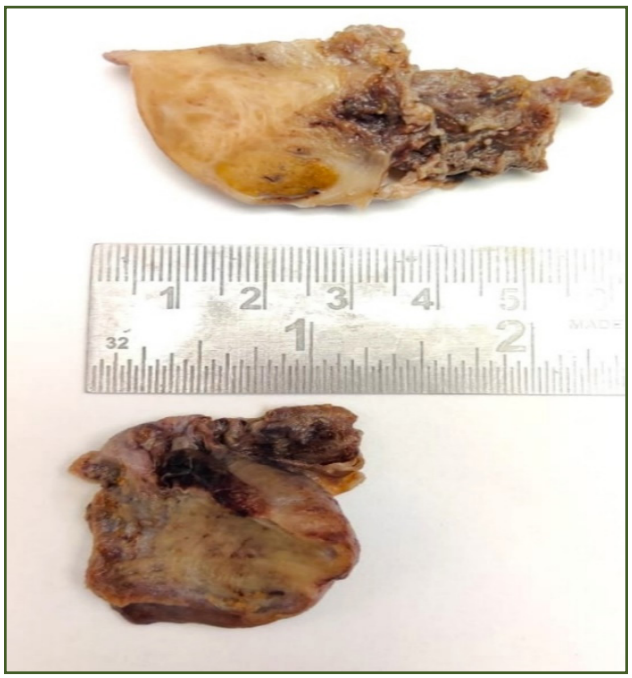

Fig. 1: Grey brown soft tissue piece (Hernial sac) with attached mass (?Nature).

Fig. 2 : Attached mass – Grey brown, external surface smooth.

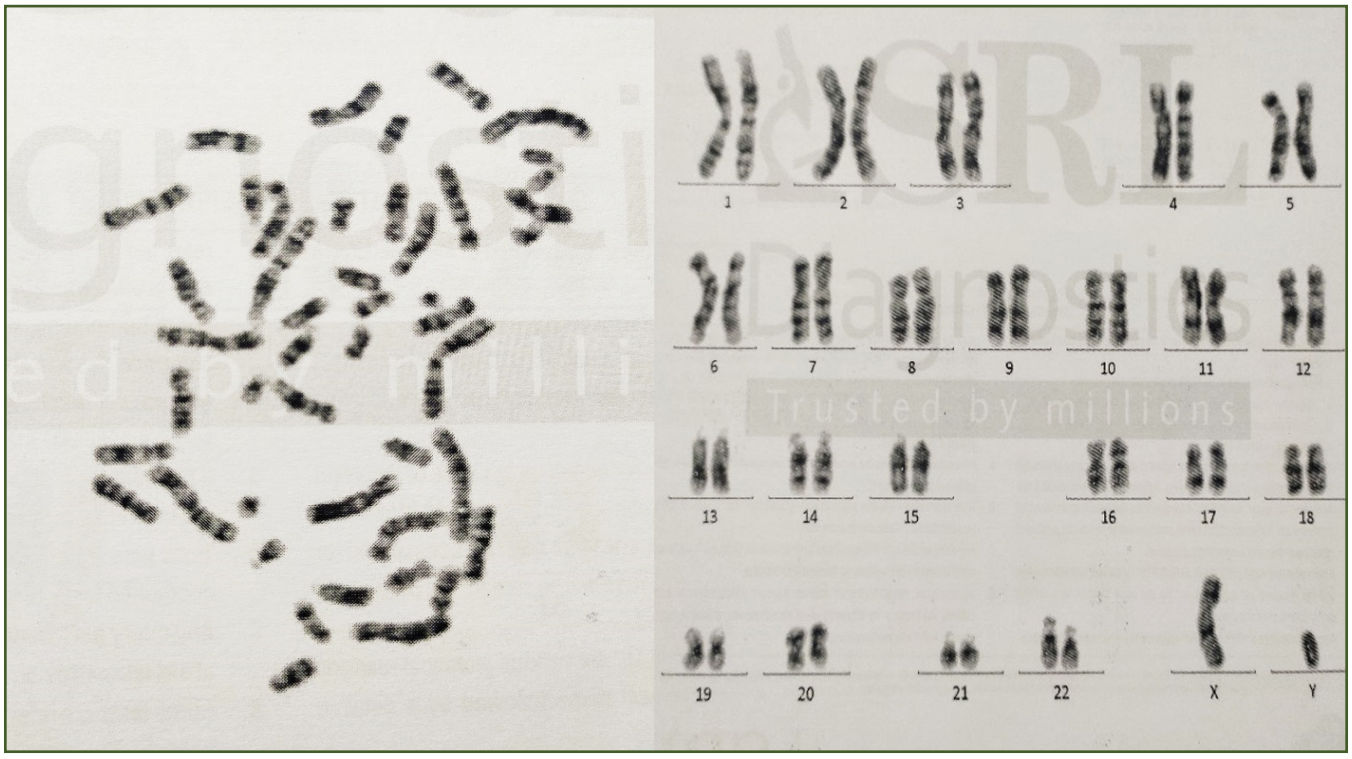

Fig. 3 : Metaphase plate and Karyotype of the subject.

uncertain. Imaging techniques (Ultrasonography, MRI, CT) and hormonal assays helps to reach the diagnosis, whereas the confirmation is done by karyotype genetic mapping, as done in the present case. Another topic at hand is the moment the orchiectomy should be performed, since the testes provide the natural levels of estrogen through the aromatizing of testosterone. Various studies show a risk of malignancy of undescended intra-abdominal testes of 3.6% at 25-year-old, and of 33% at 50-year-old. However, since hormone substitution therapy can be so easily administered nowadays, and artificial puberty is so similar to the natural puberty, there is no reason to wait and take an unnecessary chance. In addition, the last issue is whether or not to disclose the entire pathology to the patient. The family should be informed, and together with the medical staff a resolution in this regard should be found. A psychologist should always be addressed before reaching a decision, so every aspect of this situation is known and understood. As demonstrated by the present case, the 46-XY phenotypic female is almost always infertile. However, carrier females have a 50% chance of transmitting the mutated AR gene in each pregnancy. Carrier testing is advocated within the family because the disease has known familial tendencies. The present patient was the only case in her family and no carrier testing has been done in other family members.[3]

Conclusion

Testicular feminization is a rare disease that must be diagnosed and treated through close work between gynecologists, endocrinologists, geneticin's, urologists, anatomic pathologists and psychiatrists. Karyotypic analysis is the gold standard to confirm the diagnosis of testicular feminization syndrome. Bilateral laparoscopic orchiectomy is the best procedure to remove the intra-abdominal testes, in order to avoid their malignant transformation.

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Competing Interests

None declared

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