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A RARE CASE OF ANDROGEN INSENSITIVITY SYNDROME WITH BILATERAL GONADOBLASTOMA

Surgery

KEYWORDS: AIS, GONADOBLASTOMA, LAPROSCOPIC BILATERAL GONADECTOMY,MORRI'S SYNDROME, TURNER SYNDROME

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ABSTRACT

AIS is a rare inherited form of male pseudohermaphroditism that can manifest as a normal female phenotype without müllerian derivatives and absence of the upper third of the vagina.[1] The androgen insensitivity syndrome (AIS) is a disease connected with the inactivation of AR due to a mutation that inactivates male sexual differentiation, and causes a spectrum of phenotypic anomalies having as a common aspect the loss of reproductive characteristics.[2] The androgen insensitivity syndrome occurs in one out of 20,000 births and can be incomplete (various sexual ambiguities) or complete (the person appears to be a woman

INTRODUCTION

AIS is a condition that results in the partial or complete inability of the cell to respond to androgens[3] *androgenic hormones*) that stimulate or control the development and maintenance of male physiological characteristics by binding to androgen receptors.[4] The unresponsiveness of the cell to the presence of androgenic hormones can impair, or prevent, both the masculinization of male genitalia in the developing fetus, and the development of male secondary sexual characteristics at puberty, though it does not significantly impair the development of female genital or sexual characteristics in females and males with the condition.

CASE STUDY

A 27-year-old patient is admitted for primary amenorrhea. The clinical examination shows a female phenotype: the breasts are normally developed, but pubic and axillary hairs are absent, the labia are small and hypoplastic, the urinary meatus is normally inserted, and the vulva is unpigmented. The gynecological examination reveals that the hymen is present, the vagina is 1.5 cm in length, while the uterus is absent. At Endocrinology, the levels of gonadotropins were measured and found normal (FSH 69.88 mU/mL, LH 82.84 mU/mL), the progesterone was 0.19 ng/ml, estradiol was 13 pg/ml and the testosterone was 151.4 ng/dl. The karyotype was mapped in order to differentiate the androgen insensitivity syndrome from other genetic abnormalities, like Klinefelter syndrome (46XXY), Turner syndrome (45XO), mixed gonadal dyssynergia (45XO/46XY) or tetragametic chimerism (46XX/46XY). These tests confirmed the suspected diagnosis - testicular feminization (46XY). The pelvic MRI scan revealed the lack of uterus and ovaries, hypoplastic vagina, and intra-abdominal testes. The testes were removed in order to avoid the malignant risk. We performed laparoscopic bilateral orchiectomy.

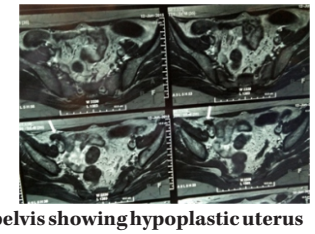

Figure 1 MRI pelvis showing hypoplastic uterus

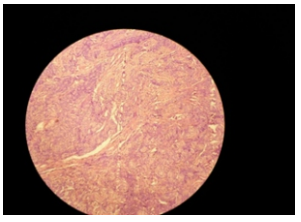

Figure 2 Histopathology report of the specimen of intra abdominal testes showing nests of dysgerminoma-like germ cells and sex cord derivatives resemble immature Sertoli and granulosa cells

CONCLUSIONS

In complementation to hormonal tests and cytogenetic techniques, laparoscopic gonadectomy is required to complete the diagnostic work up for AIS as it also adds a final therapeutic approach with low risk and huge benefit. Since laparoscopy is now a well-tolerated and widely accepted gold standard, it should be included in routine management for patients with AIS. Risk of malignancy in PAIS should be investigated in larger cohort of these patients. [5] If a primary amenorrhea is checked, the karyo-typing is compulsory. If a Morris's syndrome is suspected, it is of critical importance to find the rudimentary male gonads (by means of MRI, pelvic echography, laparoscopy) and surgically remove them to prevent the onset of malignancies (teratoblastoma, gonadoblastoma). Once the considered disease has been identified, a continuous psychological help can be considered useful for the patient and the family. Testicular feminization is a rare disease that must be diagnosed and treated through close work between gynecologists, endocrinologists, geneticians, urologists, and psychiatrists. Gonadoblastoma can be evident even at an early age in streak gonads with Y mosaicism and may be bilateral [6]. If a Y chromosome or the possibility of a Y chromosome cannot be excluded, gonadectomy should be performed because of the risk of malignancy.[7,8]

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