A RARE CASE OF ANDROGEN INSENSITIVITY SYNDROME WITH BILATERAL GONADOBLASTOMA

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 Turner syndrome (45XO), mixed gonadal dyssynergia (45XO/46XY) or the possible 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 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].

REFERENCES: