ISGE
GYNECOLOGICAL ENDOCRINOLOGY
THE 18TH WORLD CONGRESS
7-10 MARCH 2018 FLORENCE, ITALY

Mixed Germ Cell Tumor in Androgen Insensitivity Syndrome: A Case Report

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INTRODUCTION: Androgen insensitivity syndrome is a disorder of sexual development characterized by female phenotype with 46XY karyotype. Pathogenesis involves mutation in the androgen receptor gene located on the proximal long arm of the X chromosome at Xq11-12, which results in unresponsiveness of the target organs to androgen stimulation. Most present with primary amenorrhea; however, 1.1 % of patients present with an inguinal mass. This report presents the case of a phenotypically female adolescent diagnosed with AIS associated with mixed germ cell tumor.

CASE OUTLINE: A 15-year-old female looking adolescent presented with a gradually enlarging abdominopelvic mass. Thelarche and growth spurt occurred at 13 years of age with no development of axillary and pubic hairs. She remained amenorrheic. Breast development is at Tanner stage 2. There is a 14x14 cm solid non-movable, non-tender mass at the hypogastric area. Her labia majora and minora were well-developed and the urethral and vaginal openings were both patent. Hymen was intact with no clitoromegaly. Sonographic studies revealed absence of cervix and uterus. Instead, a 16.2 x 9.4 x 7.8 cm lobulated heterogeneous solid abdominopelvic mass and a 2.9 x 2.0 x 1.5 cm solid mass at the right adnexal area were seen. Laboratory tests showed elevated serum levels of luteinizing hormone, elevated follicle stimulating hormone, normal testosterone and low estradiol. Karyotyping showed 46XY. Emergency surgical resection of the mass was done due to its obstructive effects. Histopathologic examination revealed mixed germ cell tumor – seminoma and yolk sac. Post-operative chemotherapy was instituted.

CONCLUSION: The diagnosis of AIS is made based on the physical examination findings, endocrine profiling, imaging studies and karyotyping. Age and location increases the risk of malignant transformation. Cryptorchidism carries a 10% risk of developing testicular tumor and the risk of malignancy for intra-abdominal testis is 3.6% at 25-year-old, and 33% at 50-year-old. Hence, prophylactic gonadectomy is advocated. If early malignant transformation has occurred, gonadectomy becomes therapeutic. Germ cell tumors are reported in 2% of adult patient with AIS but is very rare during childhood and adolescent. Management of AIS involves multidisciplinary approach involving a gynecologist, oncologist, urologist, pediatrician, endocrinologist, geneticist, pathologist and psychiatrist.

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