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

P25. A case of 46,XY gonadal dysgenesis with remaining epididymis mimicking ovotesticular disorder of sexual development

E S Ferreira Filho (BR) [1], G d Maffazioli (BR) [2], C P Lopes (BR) [3], M I Aun (BR) [4], S A Hayashida (BR) [5], E C Baracat (BR) [6], G A Maciel (BR) [7]

CONTEXT: Complete gonadal dysgenesis 46,XY is characterized by the absence of SRY gene activity. Therefore, normal testes are not developed, and fibrous streak gonads are formed instead. OBJECTIVE: The present study aims to show a rare clinical case in which remaining epididymis and ovarian stroma were found in the gonads. METHODS: This is a patient-consented case report of a single patient. PATIENT, INTERVENTIONS AND RESULTS: Thirty-three-years-old patient with female phenotype referred primary amenorrhea. Thelarche and pubarche happened by the age of 13, and her menarche was reached by the age of 18 after oral contraceptive pill use. At physical exam, she was at Tanner stage B5 P5, with an hypotrophic cervix and normal but atrophic vagina. Laboratory workup showed an elevated FSH level (57 IU/L), low estradiol (<15 pg/mL) and testosterone (<12 ng/dL), normal levels of TSH (2.09 mUI/L) and prolactin (6.1 ng/mL) and karyotype 46, XY. Pelvic ultrasound showed and small but normal uterus (13.6 cm^3) with a corpus/cervix index < 1. She underwent to salpingo-oophorectomy due to her Y karyotype. Gonadal histologic findings were a fragment of primitive gonad exhibiting focus of ovarian stroma and rudimentary epididymis, presence of remaining müllerian structures and absence of testicular parenchyma. CONCLUSIONS AND FINAL CONSIDERATIONS: At eight weeks of intra-uterine age, proximal portion of mesonephric ducts are contorted to form epididymis. In female fetuses, mesonephric ducts completely disappear remaining only some non-functional structures. In the absence of SRY activity, there is no production of testicular determining factor, which confers a dysgenetic aspect to the gonads. Ovarian stroma is then formed by conjunctive tissue and interstitial cells derived by embryonic mesenchyme. Ovarian development occurs due to the presence of X chromosome and autosomal genes. This case identifies the rare presence of remaining epididymis and ovarian stroma in a complete 46XY gonadal dysgenesis.

[1] Hospital das Clínicas da Faculdade de Medicina da Universidade de São Paulo, [2] Hospital das Clínicas da Faculdade de Medicina da Universidade de São Paulo, [3] Hospital das Clínicas da Faculdade de Medicina da Universidade de São Paulo, [4] Hospital das Clínicas da Faculdade de Medicina da Universidade de São Paulo, [5] Hospital das Clínicas da Faculdade de Medicina da Universidade de São Paulo, [6] Hospital das Clínicas da Faculdade de Medicina da Universidade de São Paulo, [7] Hospital das Clínicas da Faculdade de Medicina da Universidade de São Paulo