SUCCESSFUL PREGNANCY AND DELIVERY IN A PATIENT WITH XY GONADAL DYSGENESISMS

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A 24-years-old female referred to our outpatient service with primary amenorrhea and sexual infantilism. She had female phenotype, eumetabolic state and eunochonoid proportion. The external genitalia were completely female and Mülleran structures were normal. A transvaginal ultrasound showed a hypoplastic uterus. Breasts were bilateral hypoplastic and the pituitary gland was anatomically normal. Hormonal tests revealed a hypergonadotropic hypogonadism. The patient started hormonal replacement therapy, with incremental doses of estradiol hemihydrate for 21 days with an addition of nomegestrol acetate 2,5 mg in the last 12 days followed by 7 days break. Development of the uterus was determined and improvement in bone mass occurred. Cytogenetic analyses showed a 46, XY karyotype. Screening of the SRY coding region identified a novel rearrangement, resulting in an insertion of adenine at position 331. The patient underwent laparoscopic bilateral gonadectomy due to risk of malignancy. At the age of 29 she underwent oocyte donation and IVF. The patient continued the HRT until the 12 week of pregnancy. The antenatal period was uneventful. The patient was admitted to our Institution at 39+6 weeks after spontaneous onset of labor. At 40 weeks she delivered a boy weighing at birth 3520 g with Apgar score of 9 and 9 at 1 and 5 minutes; artery blood pH was 7,26. Post-partum period was regular. Pure gonadal dysgenesis, also known as Swyer syndrome, is a sex-reversal disorder resulting from embryonic testicular regression sequences especially during the first few weeks of fetal life. Although patients are infertile, pregnancies have occurred in these women via donated fertilized oocytes. In literature there are 12 cases of successful pregnancy. Our case is the second report of successful vaginal birth at term. The patient reached the 40 weeks of pregnancy without complications and with spontaneous onset of labor, uneventful vaginal delivery and normal postpartum period.