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Non-obvious diagnosis and breast development in pure gonadal dysgenesis

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ABSTRACT

Pure gonadal dysgenesis is a situation when the karyotype is 46, XY, but for various reasons there is a disorder of differentiation of Wolffian and Mullerian structures and in consequence the phenotype is female. It is known that abdominal gonads and the presence of Y chromosome allow to qualify this condition as a high risk of tumor. In most cases breast development is limited because of lack or low level of estrogen. A 27-year-old patient with differences of sexual development (DSD), was admitted to the Department of Endocrinological Gynecology for a control examination. In the history: dysgerminoma, primary amenorrhea and ambiguous karyotype. The patient has not taken hormonal replacement therapy. The breast development is Tanner stage V.

Key words: pure gonadal dysgenesis; breast development; differences of sexual development

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We present a 27-year-old patient with differences of sexual development (DSD), who was admitted to the Department of Endocrinological Gynecology for a control examination. The medical documentation showed that at the age of 13 the patient went through a laparotomy due to an accidentally detected tumor with dimensions 16.5 x 12 x 11 cm that filled the entire pelvis minor. The tumor was completely removed including the left ovary. Histopathology revealed dysgerminoma. 1.5 year later the patient reported to the Department of Pediatric Endocrinology due to primary amenorrhea. On physical examination the patient was of tall stature, female phenotype, female external genitalia and Tanner stage III pubertal development. Patient was reared as female. Hormonal tests showed hypergonadotropic hypogonadism and normal level of testosterone. Ultrasound examination demonstrated prepubertal uterus. The cytogenetic examination was administered with result 46, X, +mar. Because of clinical picture, dysgerminoma in the past and ambiguous karyotype it was decided to verify cytogenetic results in another genetic laboratory. Finally, the karyotype: ish Yp11.3 (SRY+),Xp11.1q11.1(DXZ1+) was confirmed by fluorescence in situ hybridization (FISH) using peripheral blood samples. The Magnetic resonance imaging (MRI) demonstrated structure corresponding to dysgenetic gonad on the right, without follicles typical to ovaries. In line with indications, the structure was removed by surgery. Despite the recommendations, the patient has not taken hormonal replacement therapy. The patient's breast development is Tanner stage V. Uterus ultrasound picture is shown in Figure 1. The patient's hormone levels are shown in Table 1. Patient complains of hot flushes and mood swings. The psychological condition is good.

According to the Consensus Statement from 2006 the definition of DSDs is "congenital conditions within which the development of chromosomal, gonadal, and anatomic sex is atypical" [1]. The situation when the karyotype is 46, XY, but for different reasons there is a disorder of differentiation of Wolffian and Mullerian structures and in consequence phenotype is female, is called pure gonadal dysgenesis. Frequency of this condition is approximately 5 of 100,000 newborns [2].

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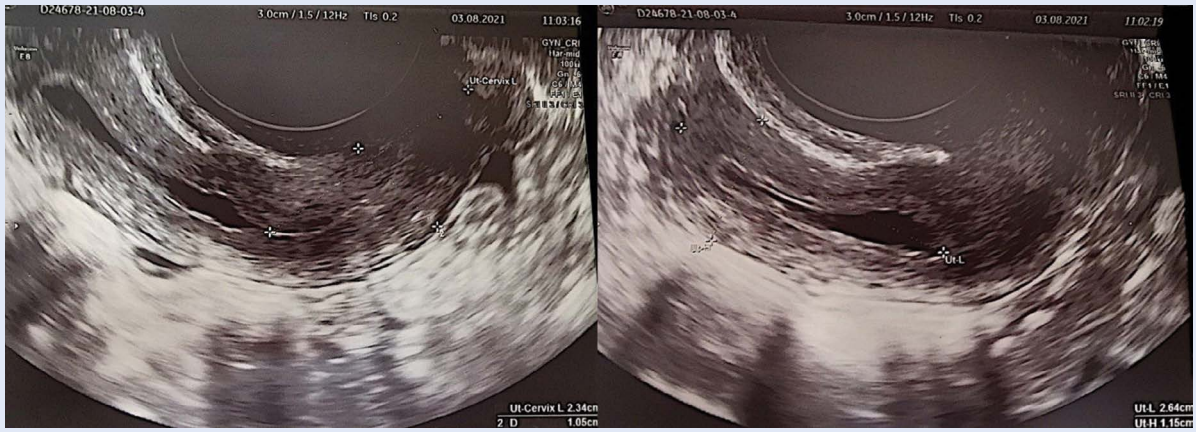

Figure 1. Current uterus ultrasound picture

Table 1. Patient's hormone results during adolescence and adulthood

	01.2010	08.2021
FSH (IU/L)	119	58.6
LH (IU/L)	29.3	10.6
Estradiol (pg/mL)	21.30	< 5.0

FSH — follicle-stimulating hormone; LH — luteinizing hormone

Estimating the risk of cancer is difficult because this DSD is rare and according to the recommendations in these cases gonadectomy should be performed as soon as possible after diagnosis, but it is known that abdominal gonads and presence of Y chromosome allow to qualify this condition as high risk of tumor [3]. In this case the situation is atypical because the first symptom of the disease was dysgerminoma which is additionally less common than gonadoblastoma [3]. Moreover, histopathology results did not raise suspicions of DSD. In most cases breast development is limited because of a lack or low level of estrogen [4]. The main aim of hormonal replacement therapy is to support development of secondary sex characteristics, including breast development, prevent osteoporosis and cardiovascular diseases [2]. It is interesting that our patient resigned from this therapy and despite that the breast development was proper. It's worth mentioning that nowadays DSDs are still being learned and the diagnosis is easier than a few years ago when the treatment of the patient started. In this case despite typical malignancy and clinical picture, diagnosis was delayed, and genetic tests results were ambiguous. The important finding is that the patient affected by the pure gonadal dysgenesis can develop proper secondary sex characteristics, even without pharmacological support. It shows how unknown DSDs still are.

Conflict of interest

All authors declare no conflict of interest.

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