Suspected burned-out ovarian germ cell tumor in Turner mosaicism: a diagnostic challenge-what is your diagnosis?

A 31-year-old nulliparous woman presented to the gynecology outpatient department with primary amenorrhea and intermittent lower abdominal pain of 10-15 days' duration. She reported a single episode of scant vaginal spotting eight years earlier. There was no history of cyclical abdominal pain, hormonal therapy, or prior evaluation for primary amenorrhea.

On general examination, she was conscious and oriented, of short stature (height 135 cm), and thinly built with a body mass index of 18.02 kg/m^2. Head and neck examination revealed a diffusely enlarged thyroid gland with multiple palpable nodules, without compressive symptoms. Cardiovascular and respiratory examinations were within normal limits.

Secondary sexual characteristics were poorly developed, with Tanner stage II breast development, absent axillary hair, and sparse pubic hair (Figure 1). Abdominal examination revealed a firm, tender abdominopelvic mass corresponding to a 26-28-week gravid uterus, predominantly cystic in consistency. External genital examination showed hypoplastic labia majora and minora, with a normal-appearing vagina.

Laboratory investigations revealed markedly elevated thyroid-stimulating hormone levels (70.8 μIU/mL) with strongly positive anti-thyroid peroxidase antibodies (>1100 IU/mL), consistent with autoimmune thyroiditis. Gonadotropins were significantly increased (follicle-stimulating hormone 127.99 mIU/mL, luteinizing hormone 47.23 mIU/mL) with low estradiol levels (17.5 pg/mL), indicating a hypergonadotropic hypogonadal state. Lactate dehydrogenase (LDH) was elevated (364 U/L), and cancer antigen-125 (CA125) was mildly raised (42.4 U/mL), while alpha-fetoprotein (2.88 ng/mL) and β-human chorionic gonadotropin (3.40 mIU/mL) were within normal limits. All other investigations, including complete blood counts, liver and renal function tests, coagulation profile, and chest radiography, were unremarkable.

Neck ultrasonography showed altered echotexture of both thyroid lobes with a multinodular goiter in the right lobe. Pelvic ultrasonography revealed a large, well-defined multiloculated cystic lesion arising from the right adnexa, with multiple internal septations. One septum measured 4-4.5 mm and showed mild vascularity. Papillary excrescences measuring 2×0.5 cm were noted inferiorly without vascularity. Internal echoes were present, with no mural nodules or calcifications (Figure 2). The right ovary was not visualized separately. The uterus was small (3.9×1.8×3.2 cm) with an ill-defined endomyometrial junction, and the left ovary was not seen.

Magnetic resonance imaging confirmed a hypoplastic uterus measuring approximately 3×6 cm, with a uterus-to-cervix ratio of 2:1 and an endometrial thickness of 2 mm. A large, thin-walled, multiloculated cystic lesion (10.6×7.2×10.2 cm) extended from the midline pelvis to the lower abdomen, reaching the level of the umbilicus. The lesion was T1 hypointense and T2 hyperintense, with irregular thickened septations (maximum thickness 5.4 mm). Some locules showed T1/FS hyperintensity, suggestive of hemorrhagic or proteinaceous content. Neither of the ovaries was visualized separately. Minimal perilesional omental edema was present, without significant lymphadenopathy or ascites. A heterogeneously enhancing tissue, extending from the uterus to the cystic lesion

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was noted, likely representing an edematous pedicle. Overall imaging features raised concern for a high-risk adnexal lesion in the setting of uterine hypoplasia (Figure 3).

Given the clinical suspicion of Turner syndrome (TS) or mosaic TS, cytogenetic evaluation was performed. Karyotyping and chromosomal microarray analysis demonstrated a 56.7-Mb deletion involving Xp22.33-Xp11.1, consistent with a structural X chromosome abnormality (Xp deletion syndrome) presenting with TS-like features, rather than classical 45,X TS (Figure 4).

Figure 1. Standing clinical photograph demonstrating short stature, underdeveloped secondary sexual characteristics, and diffuse neck enlargement

On the third day of admission, the patient developed sudden, severe abdominal pain associated with vomiting, tachycardia, and altered sensorium. In view of suspected adnexal torsion, she underwent emergency staging laparotomy under general anesthesia, despite being high-risk due to markedly elevated TSH levels. Preoperative imaging had demonstrated a large complex adnexal mass with thickened septations and papillary excrescences, accompanied by elevated LDH and mildly raised CA125 levels, raising concern for an underlying ovarian malignancy or germ cell tumor. Given the diagnostic uncertainty and the increased risk of germ cell tumors in patients with an Xp deletion syndrome and Turner phenotype, comprehensive surgical staging was performed to ensure appropriate oncologic management and avoid the need for a second procedure.

Answer

Surgery revealed a large right adnexal cystic mass measuring approximately 12×10×11 cm, with omentum adherent to its superior surface. The pedicle showed three complete twists, one of which appeared chronic with dense adhesions and was not amenable to detorsion. The uterus was hypoplastic, the left ovary was a streak gonad, and the right adnexa was incorporated into the mass (Figure 5). Minimal blood-tinged fluid was present in the pouch of Douglas and was sent for cytology. After partial detorsion, a right salpingo-oophorectomy was performed, followed by total abdominal hysterectomy, left salpingo-oophorectomy, bilateral pelvic lymph node dissection, and omental and peritoneal biopsies.

Gross examination showed a cystic right adnexal mass measuring 12.5×11.5 cm with a congested external surface and intact capsule. The cut surface was multiloculated, containing approximately 100 mL of hemorrhagic fluid, with no identifiable viable areas. The uterus and cervix were hypoplastic (6×3.5×1 cm). The left ovary was a streak gonad measuring 0.8×0.8 cm.

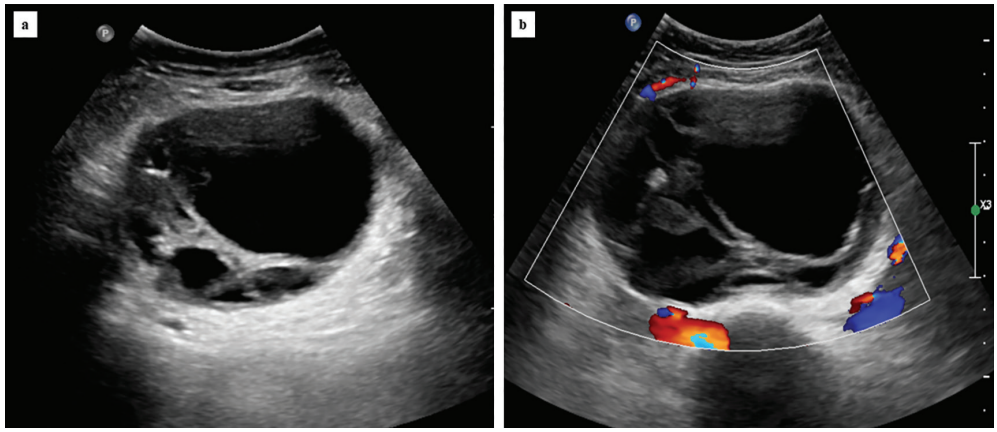

Figure 2. (a) Axial ultrasound image-large thick-walled multiloculated cystic lesion with thick internal separations; (b) On colour Doppler, minimal uptake is noted only in the wall of the lesion

Microscopically, the right adnexal specimen, including the cyst wall and residual streak gonadal tissue, was extensively sampled (20 tissue blocks). Sections revealed a fibrocollagenous cyst wall with extensive hemorrhagic necrosis and granulation tissue formation, without viable epithelial lining or identifiable ovarian parenchyma. The left streak gonad was entirely submitted for histopathological examination (2 tissue blocks) and consisted of fibrocollagenous and adipose tissue, with no follicles or ovarian stroma. Both fallopian tubes showed vascular congestion with poorly developed plicae. The endometrium was thin and inactive, lacking cyclical changes. Pelvic lymph nodes, omental and peritoneal biopsies were unremarkable, and peritoneal fluid cytology was negative for malignancy. Owing to extensive hemorrhagic ischemic necrosis with complete loss of viable tissue, the exact nature of the adnexal cyst could not be definitively established (Figure 6). Immunohistochemical studies were not performed, as the absence of viable tumor cells rendered them non-contributory. TS results from the complete or partial loss of one sex chromosome and affects approximately 1 in 2,500-3,000 live female births. Its clinical spectrum is broad, encompassing short stature, ovarian dysfunction, and variable neurocognitive features (1,2). Cytogenetically, TS is heterogeneous and only about 40% of individuals exhibit a classic 45, X karyotype, while

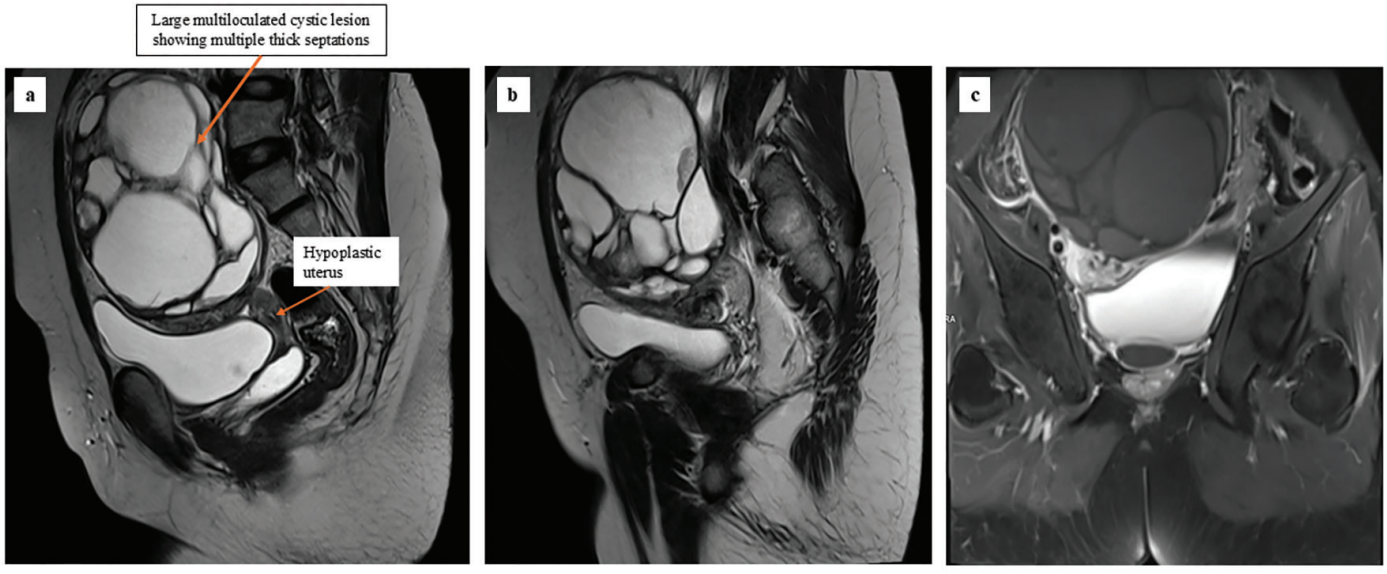

Figure 3. (a) Sagittal T2W image shows a hypoplastic uterus with preserved zonal anatomy. The right adnexal thick-walled multiloculated lesion is noted abutting the fundus and indenting the dome of the urinary bladder; (b) Right parasagittal T2W image shows edematous, thickened pedicle of the ovarian lesion; (c) Post-contrast T1FS coronal image. A thickened pedicle is seen. The ovarian lesion shows smooth wall enhancement. There is no enhancement of septations

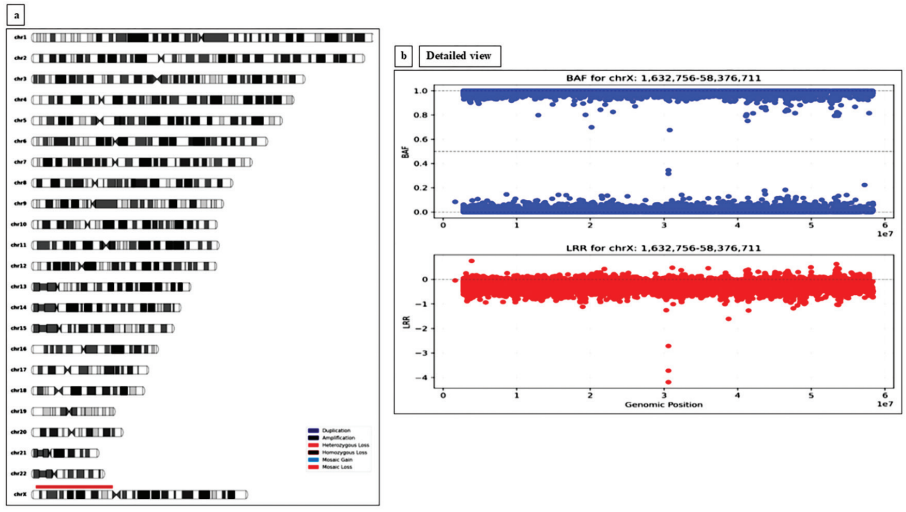

Figure 4. (a) Karyoview; (b) Detailed view of Xp22.33p11.1

the remainder show mosaicism or structural abnormalities such as deletions, ring chromosomes, or isochromosomes (2). Normal ovarian development and fertility depend on the structural and functional integrity of the X chromosome. In TS, accelerated germ cell apoptosis begins early in fetal life, leading to depletion of the primordial follicle pool, impaired folliculogenesis, and streak gonad formation (3). This process has been attributed to abnormal meiotic pairing of the X chromosome, disrupted oocyte–granulosa cell interactions, and haploinsufficiency of key X-linked genes involved in ovarian maintenance, including *BMP15* and *PGRMC1* (3,4). Clinically, most individuals develop primary ovarian insufficiency, diagnosed by amenorrhea with elevated gonadotropins reflecting hypergonadotropic hypogonadism, although a subset demonstrates partial pubertal development, menarche, or even spontaneous fertility (3).

Diagnosis of TS relies primarily on conventional karyotyping, typically assessing 15-30 metaphases, with expanded analysis recommended when mosaicism is suspected (2,3). However, standard karyotype and chromosomal microarray techniques may fail to detect low-level or tissue-restricted Y-chromosome mosaicism, referred to as "hidden Y mosaicism," for which molecular methods and fluorescence *in situ* hybridization (FISH) may be required (5,6). In the present case, additional molecular testing, including FISH or polymerase chain reaction-based analysis for Y-chromosome sequences, could not be performed because of financial constraints. Consequently, the presence of occult low-level or tissue-restricted Y-chromosome mosaicism cannot be completely excluded.

Furthermore, the reported prevalence of Y-chromosome material in TS varies widely (4-61%), depending on the detection method, and carries a clinically significant risk of 10-30% for gonadoblastoma (7).

Individuals with TS and dysgenetic gonads are therefore at increased risk for gonadal germ cell tumors. Gonadal dysgenesis reflects abnormal fetal gonadal development resulting from sex chromosome abnormalities or mutations in genes that regulate urogenital ridge formation and sex determination (8). Among germ cell tumors associated with dysgenetic gonads, gonadoblastoma is the most characteristic lesion. It is a borderline neoplasm composed of germ cell and sex cord elements, frequently bilateral, and capable of malignant transformation, most commonly into dysgerminoma or seminoma, in up to one-third of cases (7,9). Dysgerminoma, the ovarian counterpart of testicular seminoma, typically presents in adolescents and young women and is often associated with elevated serum LDH levels, which may aid in diagnosis and follow-up (10-12). Moreover, the mildly

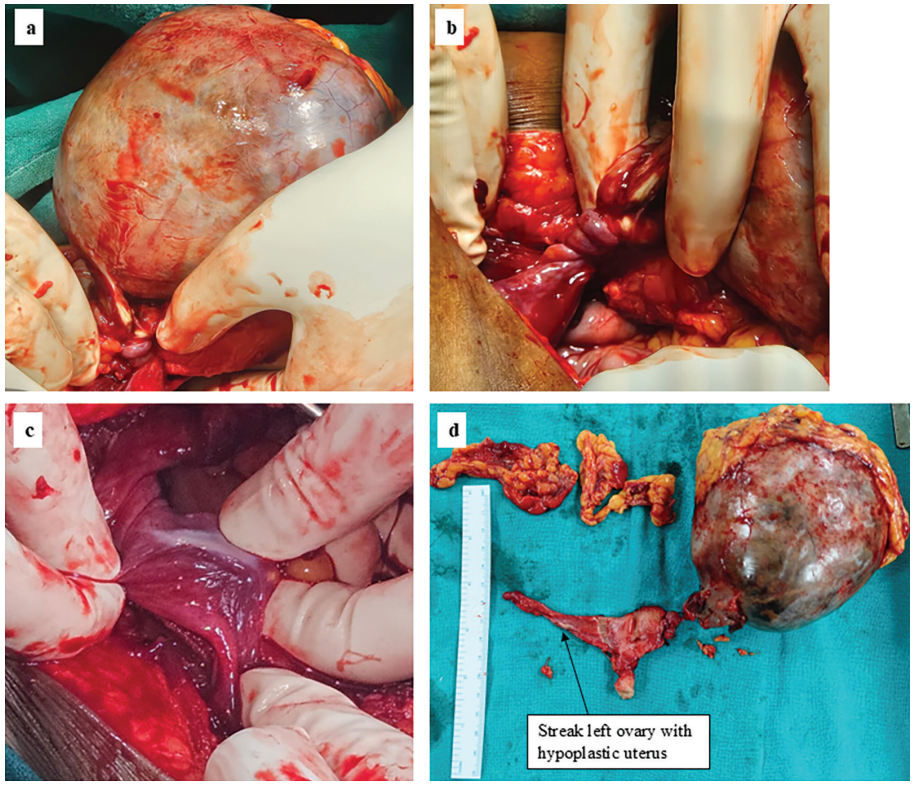

Figure 5. (a, b) Intraoperative images showing a large right adnexal mass with multiple twists in the pedicle; (c) Left streak ovary with fibrous appearance; (d) Post-hysterectomy specimen showing a hypoplastic uterus with a streak left ovary and a large right adnexal cyst with lymph nodes and omental biopsy

elevated serum CA125 level observed in the present case may be attributable to chronic adnexal torsion, inflammation, and extensive hemorrhagic necrosis rather than malignancy alone, as has been reported in previous studies (13).

In the present case, Y-chromosome material was not identified on chromosomal microarray analysis. Nevertheless, given the dysgenetic gonadal phenotype and tumor characteristics, the possibility of occult or low-level Y mosaicism cannot be definitively excluded. An additional diagnostic consideration is the phenomenon of spontaneous tumor regression. "Burned-out" germ cell tumors are well documented in the testis, particularly in seminoma and embryonal carcinoma, where the primary lesion undergoes regression, leaving fibrotic or necrotic remnants, sometimes in the presence of metastatic disease (14,15). In contrast, spontaneous regression of ovarian germ cell tumors is exceedingly rare, and true burned-out ovarian dysgerminoma remains poorly characterized (16).

In this patient, chronic adnexal torsion resulted in extensive hemorrhagic necrosis with complete loss of viable tumor tissue, accompanied by elevated serum LDH levels and bilateral dysgenetic gonads in the setting of Xp deletion syndrome with a Turner phenotype. While ischemic necrosis from long-standing torsion is a plausible explanation, the overall clinicoradiological and biochemical profile raises the possibility of a burned-out ovarian germ cell tumor. Although definitive confirmation is limited by the absence of residual or metastatic disease, this presentation broadens the differential diagnosis of adnexal masses in patients with a TS phenotype.

This case also has important implications for endocrinologists, pediatricians, gynecologists, and pathologists involved in the care of individuals with TS and disorders of sex development. Patients with features of TS presenting with primary amenorrhea, hypergonadotropic hypogonadism, dysgenetic gonads, or adnexal masses should undergo comprehensive genetic evaluation, including assessment for occult Y-chromosomal material when clinically indicated, as its presence significantly increases the risk of gonadoblastoma and other germ cell tumors. Although no Y-chromosome material was detected in the present case, the possibility of low-level or tissue-restricted mosaicism cannot be completely excluded. This highlights the importance of individualized risk stratification, timely consideration of prophylactic gonadectomy in high-

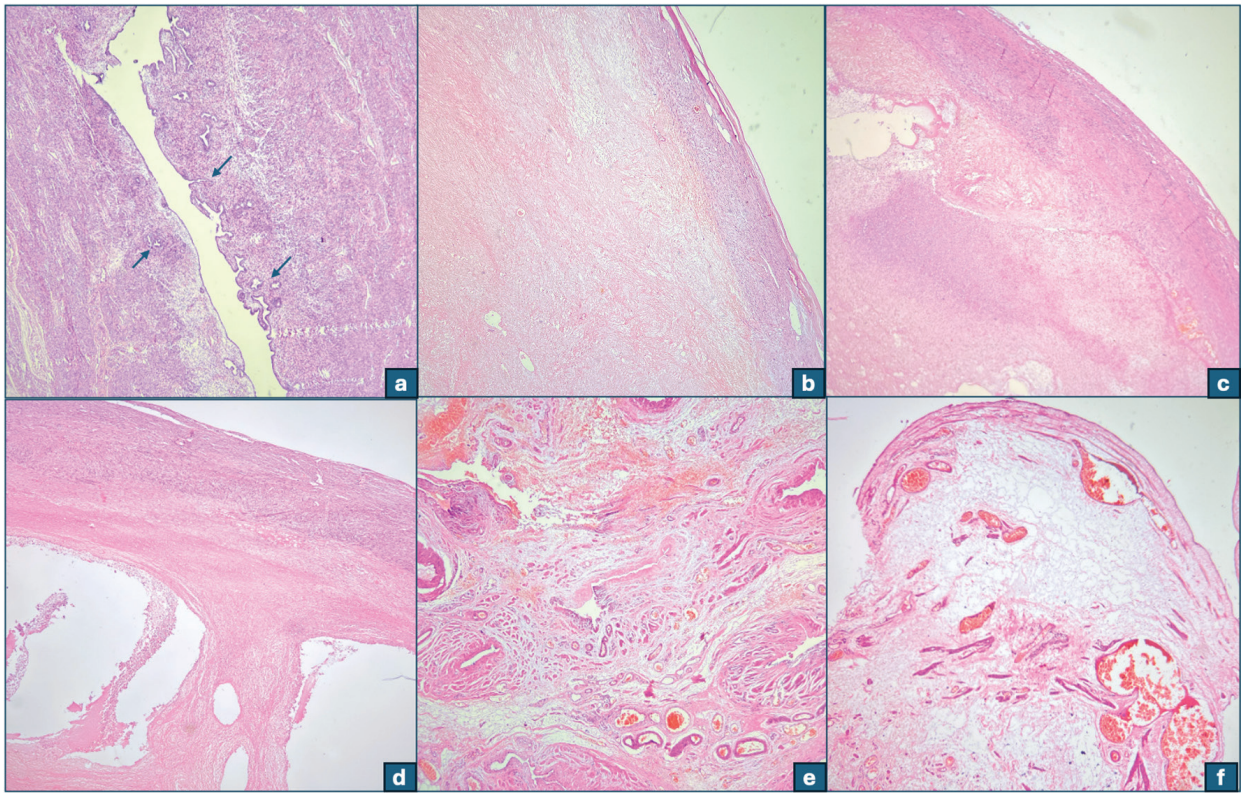

Figure 6. (a) Hypoplastic uterus showing poorly developed endometrial lining (arrow) with scant, inactive glands (H&E, 40x); (b-d) Right adnexal cyst demonstrating extensive haemorrhagic necrosis with loss of viable epithelial lining, precluding definitive characterization of the cyst [(H&E, 40x (b),100x (c)]; (e, f) Left and right streak gonads composed predominantly of fibrocollagenous stroma with variably sized blood vessels with complete absence of ovarian follicles or parenchyma (H&E, 100x)

H&E: Hematoxylin and eosin

risk patients, and multidisciplinary management involving gynecology, endocrinology, pathology, genetics, and pediatric specialists to optimize diagnosis, surgical decision-making, long-term endocrine care, and surveillance.

This case illustrates the diagnostic complexity of evaluating ovarian and adnexal masses in patients with Xp deletion syndrome presenting with a Turner phenotype and gonadal dysgenesis. Even in the absence of detectable Y-chromosome material on standard testing, low-level or tissue-restricted mosaicism cannot be excluded. The coexistence of a large adnexal mass, elevated LDH, and extensive hemorrhagic necrosis without viable tumor tissue raises suspicion of a burned-out ovarian germ cell tumor, an entity well recognized in the testis but rarely described in the ovary. This report expands the limited literature on atypical presentations of germ cell tumors in patients with Xp deletion syndrome and a Turner phenotype. Furthermore, this case highlights the need for heightened clinical vigilance, thorough clinicopathologic correlation, individualized surgical management, and long-term surveillance in patients with dysgenetic gonads.

Ethics

Informed Consent: *Written informed consent was obtained from the patient both for participation in the study and for publication of the relevant data.*

Footnotes

Conflict of Interest: *No conflict of interest is declared by the authors.*

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