To the Editor: In June 2013, a 17-year-old patient was admitted to our hospital for a pelvic mass and primary amenorrhea. The patient’s cousin had a similar clinical presentation without a definite diagnosis. Physical examination revealed a phenotypic female with a slender body, poor mammary gland development, an infantile vulva, no pubic hair, and a normal-appearing vaginal orifice and vestibule. A hard and solid abdominal mass, approximately 12 cm in diameter, was found. The vagina was approximately 3.0 cm in length with a blind end measured by cotton swabs. The human chorionic gonadotropin (HCG) (40.64 mU/ml) and lactate dehydrogenase (LDH) (431 U/ml) levels were elevated. The follicle-stimulating hormone (111 mU/ml) and leutinizing hormone (27.8 mU/ml) levels were also elevated, while the E2, T, and A levels were below the reference values. The diurnal cortisol and adrenocorticotropic hormone (ACTH) levels were normal. Chromosome karyotype analysis was then done and revealed a result of 46XY. Pelvic computed tomography scan revealed multiple masses posterior to the uterus and involving the retroperitoneal abdominal aorta to the level of the left renal veins. Hysteroscopy was performed and showed the vagina to be 5 cm in length with a blind end apex, and a 2 cm × 2 cm cervical structure without an orifice was visible. An open exploration was performed and detected a 12 cm × 10 cm × 9 cm solid pelvic mass with a purple capsule and an abundance of surface blood vessels, with poorly developed fallopian tubes. A strip-like rudimentary uterus with an atrophic right ovary was present on the right side of the mass, suggesting the mass to be the left ovary. Several masses with smooth surfaces, complete capsules, and a grey color were adjacent to the left side of the abdominal aorta to the level of the renal arteriovenous connection, the largest diameter of which was 6 cm × 5 cm × 4 cm. Intraoperatively, the masses were removed first. A frozen section could not be performed because the patient had hepatitis. Based on the preoperative assessment and intraoperative findings, we considered the mass to be a malignancy, thus an abdominal aorta lymph node dissection and omental biopsy were performed. The rudimentary uterus was also removed to prevent cervical atresia due to a poorly developed cervix.

Pathologic evaluation reported the mass to be a dysgerminoma and gonadoblastoma with the following marker profile: CD117 (+), CEA (‐), Desmin (‐), GFAP (‐), PLAP (+), S-100 (focal +), hCG (‐) and AFP (‐). Endometrium was in the proliferative phase.

Gonadoblastoma components were visible in the right ovarian stroma. Metastases to the abdominal aorta lymph nodes were detected. The postoperative diagnosis was a Stage IIIc left ovarian dysgerminoma and gonadoblastoma. Ten days postoperatively, the HCG and LDH levels were normal. Four cycles of BEP chemotherapy were completed with no evidence of tumor recurrence after 3 years. The patient also got hormone replacement therapy.

Through this case and review of the literature, we conclude a diagnostic flow chart to identify this kind of patients [Figure 1]. (1) Gonadal dysgenesis should be contemplated in patients who have primary amenorrhea, and chromosomal analysis and hormone levels should be performed. (2) Swyer syndrome should be included in the differential diagnosis of complete androgen insensitivity syndrome (CAIS) and 17α-hydroxylase deficiency syndrome, which also present with a 46XY karyotype. Blood pressure should be checked.

Figure 1: Diagnostic flow chart of primary amenorrhea patients.
be recorded, and potassium, ACTH, and diurnal cortisol levels should be measured. 17α-hydroxylase deficiency syndrome will be characterized by hypertension and hypokalemia. CAIS is usually accompanied with elevated estrogen and androgen level. (3) A resection of the gonads should be performed as soon as possible before metastasis, and preferably, before tumor formation.[1] (4) Postoperative BEP chemotherapy is effective auxiliary therapy. (5) A normal-appearing uterus and cervix can be preserved. The development of secondary sexual characteristics and pregnancy can be achieved through HRT and assisted reproductive techniques.[2]

Declaration of patient consent
The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest
There are no conflicts of interest.

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