Short Communication

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A Case of Male Pseudohermaphroditism Combined with Dysgerminoma of Ovary

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A 17-year-old Tibetan patient with a social gender of female presented to the hospital with a history of cyclic lower abdominal pain for one year and exacerbation for one month. Upon admission, she was diagnosed with uterine fibroids. The patient has poor breast development, normal external genital development, unobstructed vagina, no previous menstruation, unmarried, and has a history of sexual activity. The patient experienced periodic lower abdominal pain and discomfort without obvious cause 1 year ago, lasting for 1 day each time with intervals ranging from 15-30 days. The patient was diagnosed with uterine fibroids by multiple examinations, and laboratory tests found that human chorionic gonadotropin (HCG) was 121.82mIU/ml, testosterone was 1.07ng/ml, and carbohydrate antigen CA-125 was 43.4U/ml. The ultrasonography showed the uterine body (32mm × 24mm × 20mm), endometrium (3mm), multiple mixed echo masses in the pelvic cavity, the larger one (113mm× 80mm), and star-dot blood flow signals. Both ovaries were not detected. MRI showed a huge pelvic mass, with uneven light-moderate enhancement on enhanced scans. The uterus is compressed and its volume has shrunk. The patient underwent a total hysterectomy via the abdomen + bilateral salpingo-oophorectomy + bilateral pelvic lymph node dissection + para-aortic lymphadenectomy + omentectomy + appendectomy + pelvic adhesion laxity. During the operation, a solid mass of about 100mm × 100mm was found in the plain umbilical area, which came from the left adnexa without adhesion to the surrounding tissue.

A solid mass about 120mm× 130mm in size was found in the pelvic cavity, which came from the right adnexa and adhered to the intestinal tube. After separation and adhesion, there were grains and brittle tissues on the surface of the intestinal tube and peritoneum. The uterus was small and flat, about 30mm×30mm× 20mm in size. Intraoperative diagnosis of bilateral ovarian dysgerminoma, infantile uterus, Swyer syndrome. It was confirmed by pathology as ovarian dysgerminoma (IIIc, T3NoMo). Chromosome examination results: Karyotype 46XY.

In this case, the patient has poor breast development and no previous menstruation. The genitals are not completely masculinized, and the patient has a certain degree of developed female reproductive organs. Laboratory examination found that testosterone levels have increased. Chromosome examination results: Karyotype 46XY. The disease diagnosis is male pseudohermaphroditism. Male pseudohermaphroditismis-anautosomal recessive genetic disease with the 46XYchromosome karyotype, characterized by discoordination of chromosomes, gonads, and external sexual organs. Male pseudohermaphroditism may be caused by factors such as androgen insensitivity syndrome, 5α- reductase deficiency, anti-Müllerian hormone deficiency [1,2]. Ovarian dysgerminoma lacks characteristic clinical manifestations and may present as abdominal pain and abdominal masses. The patient was initially diagnosed with uterine fibroids. The increase of carbohydrate antigenCA-125 in the patient's laboratory examination is not specific, and the increase of HCG helps to indicate ovarian germ-cell tumor. The increase of HCG level may be the syncytial trophoblast cells in dysgerminoma produce HCG [3]. Clinically, patients with gonadal dysplasia accompanied by pelvic masses may consider the possibility of dysgerminoma.

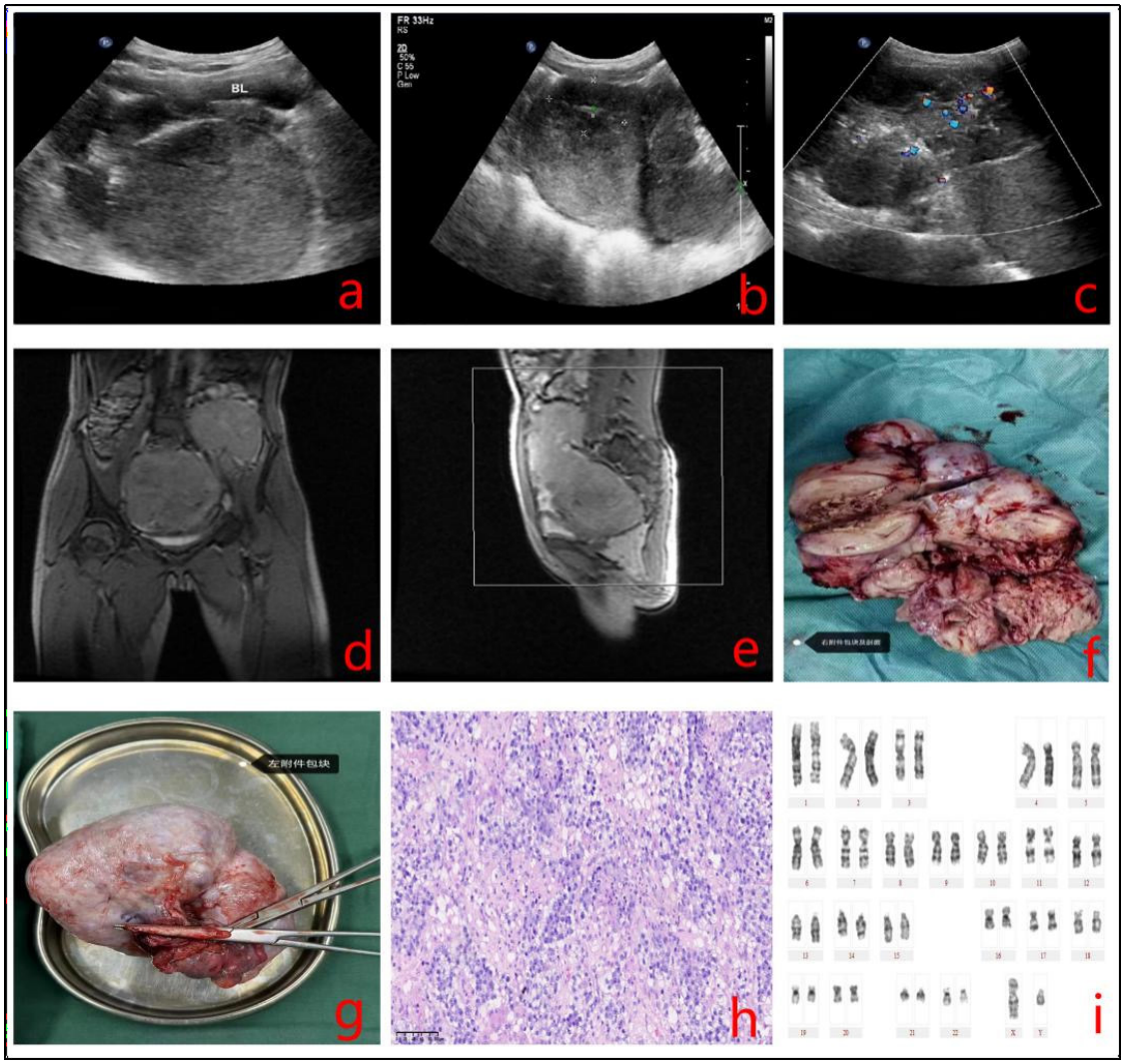

Figure 1: (a) Mixed echo mass in pelvic cavity(113mm×80mm. (b) Ultrasound examination of the uterine body (32mm ×24mm×20mm).(c) Pelvic mixed echo mass, sparse blood flow signal. (d) Coronal plane and (e) sagittal plane images show a large mixed echo mass in the pelvic cavity with uterine compression (f) Intraoperative mass in the right adnexal area. (g) Intraoperative mass in the left adnexal area. (h) Postoperative pathology: ovarian dysgerminoma. (i) Chromosome examination results: Karyotype 46XY.

The patient in this case grew up in a plateau pastoral area, had a low educational level and could not speak Chinese, which resulted in late medical treatment. The tumor was large at the time of discovery. The patient received chemotherapy with the BEP regimen and is currently doing well with follow-up.

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