

# Ovarian dysgenesis and dysgerminoma: A case report

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## Abstract

A case of a 24 year lady had presented to us with sexual infantilism, primary amenorrhoea and pain abdomen had laparotomy finding of an ovarian tumor which histologically confirmed to be dysgerminoma. In a patient with primary amenorrhoea and sexual infantilism, possibility of ovarian dysgenesis with high probability of gonadal malignancy should be kept in mind.

**Key words:** Dysgerminoma, Gonadal dysgenesis, ovarian dysgenesis and primary amenorrhoea

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## Introduction

Dysgenesis is the defective embryonic development of an organ. Ovarian dysgenesis is an abnormal ovarian development leading to rudimentary gonadal development and failure of ovarian hormone secretion. Because of the gonadal failure, patients with dysgenetic gonads will have streak ovaries and primary amenorrhoea due to lack of ovarian hormones.

## Case report

A 24 years unmarried lady presented with complaints of pain lower abdomen for 4 days, which was intermittent in nature, was mild to severe in intensity. There were no aggravating or relieving factors. She also had weakness, had never attained menarche and never sought medical advice.

On examination, she was comfortable, average built with height of 142 cm. Her secondary sexual characters were not developed. Per abdominal examination showed an oval mass of 8x6 cm in the right lumbar region which was slightly tender on palpation. Per vaginal examination was not done as patient was virgin. On digital rectal examination, uterus was not appreciated.

Ultrasonography of the abdomen and pelvis revealed a complex mass in right adnexia of 11.2 x 10.8 x 8.1 cm, with high diastolic flow suggesting malignancy. Ovaries were not visualized; uterus was small measuring 5x1.4x1 cm only. There were mild right sided hydronephrosis and small amount of ascites.

All the investigations including tumor markers Serum  $\beta$  hCG, CA 125 and  $\alpha$  fetoprotein were within normal limits. Follicular stimulating hormone level estimation could not be done at that time.

Laparotomy was done suspecting ovarian malignancy and on opening the abdomen there was hemorrhagic ascites of

about 500 ml, a right ovarian tumor of 12 x 10 cm size, which was friable and ruptured during surgery (fig 1). Uterus & other ovary were rudimentary. Other areas including liver, omentum and bowel were free of deposits and no lymph nodes were palpable. A total hysterectomy with bilateral salphingo-oophorectomy was done and surgical staging of IC was made.

Ascitic fluid cytology came out to be negative for malignant cells but Histopathology report was suggestive of Dysgerminoma. Other ovary, uterus and omentum were free of microscopic deposits.

With all above findings a final diagnosis of ovarian dysgerminoma in a dysgenetic gonad was made and patient was referred to cancer hospital for radiotherapy.

## Comment

There is a risk of development of gonadal tumor in a dysgenetic gonad. The most important part of gonadal dysgenesis is the genesis of gonadoblastoma in the presence of the Y-chromosome.<sup>1</sup> It appears that, in the X linked form, XY gonadal dysgenesis may be caused by a point deletion or mutation of a gene on the X- chromosome, which controls the gonad specific receptor for the H-Y antigen.<sup>2</sup> The absence of Sertoli cells in these patients, causing lack of androgen binding protein with deficient local concentration of androgens and consequent failure of maturation of spermatogonia, may lead to unregulated proliferation of germ cells, and hence explain the frequency of gonadal neoplasia in the 46XY female.<sup>3</sup> In a patient with 46XX karyotype, the malignant potential is probably less. In any female with gonadal dysgenesis and a Y chromosome in her karyotype, there is a need for a diligent search for the gonadectomy as soon as possible to avoid the risk of gonadal neoplasia, whether benign or malignant.<sup>3-7</sup> If the possibility of a Y chromosome cannot be excluded, gonadectomy should be performed because of the risk of malignancy.<sup>5</sup>

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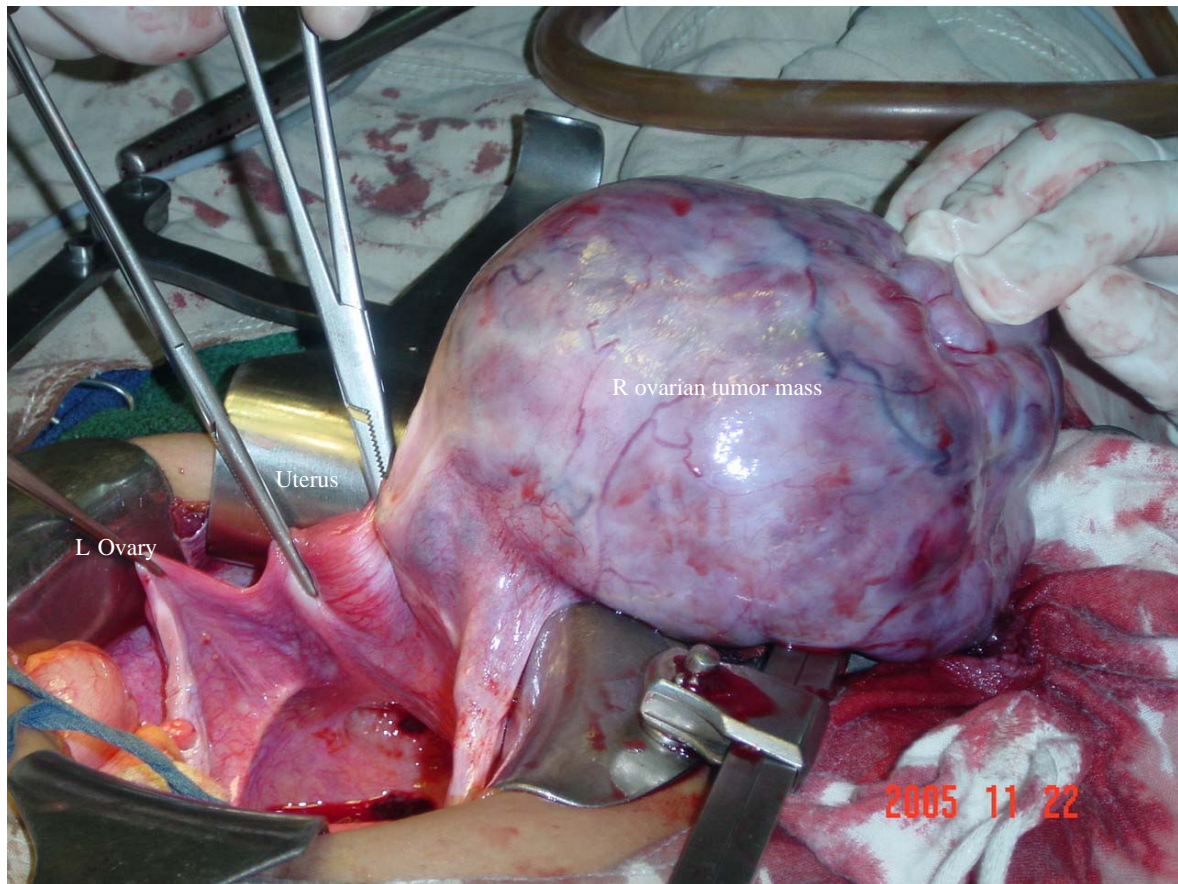


Figure 1: Right ovarian tumor with rudimentary uterus and left ovary

The high prevalence of gonadal tumors in children with mixed and pure gonadal dysgenesis warrants consideration of early, bilateral, prophylactic gonadectomy once the diagnosis is established with certainty.<sup>8,9</sup> In all patients with 45X/46XY gonadal dysgenesis and a male phenotype, gonadal biopsies should be considered as soon as the syndrome is diagnosed.<sup>10</sup> Although girls with Turner's syndrome (45X) are not at risk for malignancy, patients with feminizing testicular syndrome with XY chromosomes and patients with "mixed gonadal dysgenesis" are at risk for malignancy, and bilateral gonadectomy should be performed.

## Conclusion

Dysgerminoma in dysgenetic gonad is uncommon which further can be reduced by gonadectomy in cases of sexual infantilism with demonstrable Y chromosome sequences, that has higher propensity for malignancy.

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