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Case Report

# Mixed gonadal dysgenesis

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## Introduction

The exact incidence of mixed gonadal dysgenesis is not known. It is not very common.

## Case report

A 20 years old lady was admitted to the gynecological ward from the out patient department on 13th January, 2004. She complained of primary amenorrhoea and poorly developed secondary sex characters. There was no history of cyclical abdominal pain, hormonal intake, radiation exposure, chemotherapy, and central nervous system or gastrointestinal symptoms. There was no history of significant trauma and medical or surgical illness. Her mother had delivered her as the 11th child at the age of 44 years. She had six brothers and four sisters. All of them had attained normal puberty.

On general examination, she was 168 cm tall and weighed 70 kg with BMI of 27. There was no acanthosis, acne, hirsuitism, thyroid swelling or Cushingoid features and Turner stigmata. The breast was small with hypo-pigmented areola and no secondary mount (Tanner stage II). There was no galactorrhea. There was no axillary or pubic hair. All systems were within normal limits. The external genitalia were of female type with no pubic hair. There was no scar or swelling in the inguinal region. There was no clitoromegaly. Vaginal examination revealed an intact hymen and small rudimentary cervix.

Ultrasonography showed a rudimentary uterus and the ovaries could not be visualized. The serum FSH level was 43.6 IU/mL. Karyotyping showed a 46 XY pattern.

At laparotomy done on 16th January, 2004 after proper counseling and informed consent a rudimentary uterus, and fallopian tubes, and bilateral streak gonads were found. Bilateral gonadectomy was done. She was discharged on 21st January, 2004.

Histopathologic examination of the excised gonads showed ovarian stroma on right side (Figure 1) and epididymal tubules and rudimentary testicular tissue on the left side (Figures 2, 3).

At the followup visit after 3 weeks the patient and her relatives were counseled about the future reproductive potential. She was advised conjugated equine estrogen 0.625 ug daily continuously with 10 mg medroxyprogesterane acetate added from 21st day to 30th day of the cycle.

She was asked to come for follow up every 3 months.

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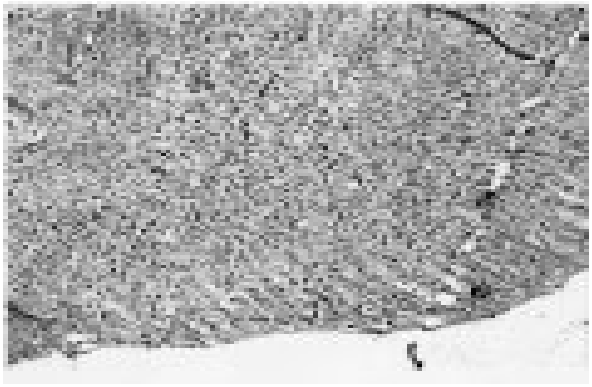

**Figure 1.** Histopathologic section from right gonad showing ovarian stroma only.

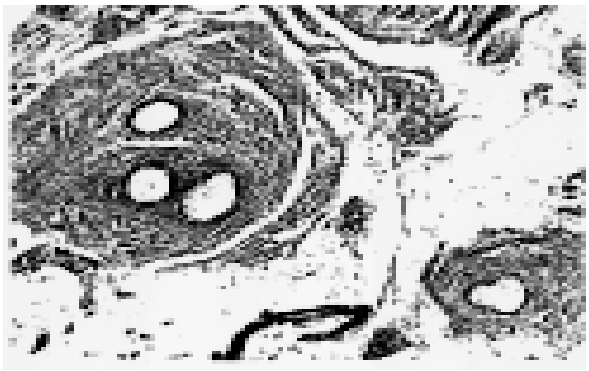

**Figure 2.** Histopathologic section from the left gonad showing epidiymal tubules.

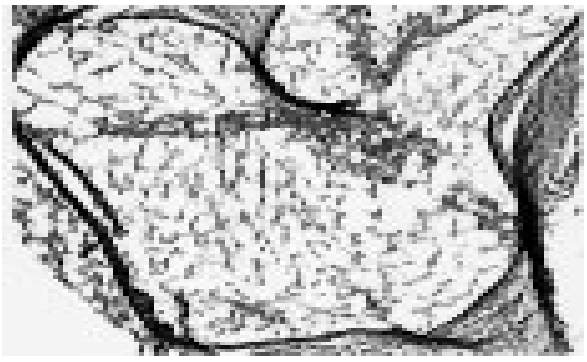

**Figure 3.** Histopathologic section from the left gonad showing rudimentary testicular tissue.

Six months after surgery the breast development had reached Tanner stage III.

## Discussion

Dysgenetic gonadal syndromes include a wide spectrum of conditions with varying gonadal development and phenotypic appearances. In this case the clinical differential diagnoses entertained were Swyer's syndrome and mixed gonadal dysgenesis. Swyer's syndrome is a pure gonadal dysgenesis occurring in 46 XY individuals, in which condition there are streak gonads bilaterally without testicular tissue [1]. A streak gonad consists of fibrous ovarian stroma without follicles [2]. In mixed gonadal dysgenesis one gonad is a fibrous streak and the other is testis which is usually rudimentary. The internal genitalia include the uterus, vagina and two fallopian tubes, despite the presence of testicular tissue on one side [2].

Histopathologic examination of the excised gonads in our case showed rudimentary testicular tissue and epididymal tubules on one side and ovarian stroma on the other side. This excludes the diagnosis of Swyer's syndrome. The incidence of gonadoblastoma in a dysgenetic gonad being 25% [3], gonadectomy was done. We could not find the exact incidence of mixed gonadal dysgenesis inspite of extensive search of the literature.

### References

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