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## OBJECTIVES

The Swyer syndrome belongs to a group of pure gonadal dysgenesis. Karyotype is 46,XY. Aberrations of chromosome Y or SRY gene mutation is present in 15% to 30% of cases. These patients have high gonadotropin levels and are classified as having hypergonadotropic hypogonadism. The Swyer syndrome in the female requires close followup because of the high risk of neoplastic transformation in the dysgenetic gonads. Herein we report a case of Swyer syndrome with gonadoblastoma and dysgerminoma.

## CASE

A 22-year-old woman was admitted to the Endocrinology Outpatient Clinic. Menarche age was 15 years old. Breast development was normal during pubertal period without using any estrogen pill. After one year of regular menstrual period, irregular vaginal bleeding occurred. At the age of 18, a big mass was felt on abdomen. She had an abdominal operation for that mass and gonadoblastoma and dysgerminoma were diagnosed pathologically on the right ovary. After that pathologic diagnosis, karyotype study was performed and found as 46,XY. As the integrity of tumor capsule was destroyed, adjuvant chemotherapy was introduced. After chemotherapy, another abdominal operation was performed and gonadoblastoma was diagnosed on the left ovary. There was no laboratory examination regarding to gonadotropin and estrogen levels before the operations. SRY positivity was present on left oophorectomy material. Phenotypically female patient with 46, XY karyotype, Swyer syndrome was diagnosed. She was started estrogen/progesterone combination pill.

## RESULTS

This patient had the normal pubertal development without any delay. Because of this reason, we thought that the gonadoblastoma was hormone active most probably with estrogen producing one. Unfortunately, we couldn't demonstrate this. Patients with gonadal dysgenesis and 46,XY karyotype should be referred for bilateral gonadectomy because of the high risk of neoplastic transformation. Estrogen producing gonadoblastoma may mask gonadal dysgenesis and delay the diagnosis of this pathology.

