



# Seminoma in A Case of Testicular Feminization Syndrome: Patient Outcome After 7 Years of Follow-up

## *Testiküler Feminizasyon Sendromunda Gelişen Seminoma: Hastanın 7 Yıllık Takibi*

Yıldız GÜNEY<sup>1</sup>, Meltem NALÇA ANDRIEU<sup>2</sup>, Ayşe HİÇSÖZMEZ<sup>2</sup>, Caner AKTAŞ<sup>2</sup>, Cengiz KURTMAN<sup>2</sup>, Şaban Çakır GÖKÇE<sup>2</sup>

<sup>1</sup> Ankara Onkoloji Hastanesi, Radyasyon Onkolojisi Kliniği, ANKARA

<sup>2</sup> Ankara Üniversitesi Tıp Fakültesi, Radyasyon Onkolojisi Anabilim Dalı, ANKARA

### SUMMARY

*This is a case of a young woman who had testicular feminization and developed a seminoma in an undescended testis. This patient admitted to the gynecology service with the complaint of primary amenorrhea. Cytogenetic analysis of chromosomes from blood demonstrated a 46, XY karyotype. At laparoscopy, a strict structure believed to be uterus was noted. The right gonad was found 10 x 15 mm in size, solid, fusiform, on the pelvic sidewall. There was no left gonad. The procedure was terminated following right gonodectomy. Pathology had reported the result as seminoma developed in the atrophic testis. She treated with irradiation for a total dose of 26 Gy in 2 Gy daily fractions by using 6 MV photons of linear accelerator. No recurrence has been found during the postoperative follow-up period of 7 years.*

**Key Words:** Testicular feminization syndrome, seminoma, radiotherapy.

### ÖZET

*Bu çalışmada, testiküler feminizasyon tanısı konan ve inmemiş testiste seminom gelişmiş 23 yaşında bir hasta bildirilmiştir. Kandan alınan kromozomların sitogenetik analizinde 46 XY karyotipi bulundu. Laparoskopide uterus olduğu düşünülen sert bir yapı bulundu. Sağ gonad pelvik duvar içerisinde 10 X 15 mm ebadında, fusiform, solid kitle olarak bulundu. Sol gonad yoktu. Sağ gonadektomi uygulandı. Patoloji atrofik testisten gelişen seminoma sonucu olarak rapor edildi. Hasta 6 MV foton LINAC kullanılarak günde 2 Gy fraksiyon ile toplam 26 Gy ışınılandı. Bu seminoma olgusu operasyon sonrası 7 yıl nüksetmemiş olarak takip edildi.*

**Anahtar Kelimeler:** Testiküler feminizasyon sendromu, seminoma, radyoterapi.

### INTRODUCTION

Testicular feminization (androgen insensitivity syndrome) presents in phenotypically normal women with normal breast development, normal external genitalia, absent uterus, a vagina of variable depth and scant or absent pubic and axillary hair. Most with this condition are not diagnosed until

they fail to menstruate. The undescended testes may be placed in intraabdominal, inguinal or labial and are at increased risk of malign transformation. Many types of testicular tumors have been reported in patients with testicular feminization. Seminoma followed by embryonal cell carcinoma is the most common neoplasma encountered in undescended testes. We hereby present a case of seminoma with

testicular feminization with 7 years follow-up after treatment.

### CASE REPORT

She was a 23 years-old single woman, referred to our clinic in August 1999 with the diagnose of seminoma. She admitted to the gynecology service with the complaint of primary amenorrhea. She had no family history. In the initial physical examination the patient was 1.66 m high and weighed 80 kg. The breast development of the patient was consistent with Tanner II and the female external genitalia appeared to be feminine character but no pubic hair. The pelvic ultrasonography and tomography revealed that there was no evidence of internal genitalia.

Cytogenetic analysis of chromosomes from blood demonstrated a 46, XY karyotype. Hormonal profile of the patient was as following, follicle-stimulating hormone (FSH): 25 mIU/mL; luteinizing hormone (LH): 30.1 mIU/mL; testosterone: 0.53 ng/dL; cortisol: 17.1  $\mu$ g/dL; dehydroepiandrosterone sulfate: 131  $\mu$ g/dL. The hemogram profile, biochemistry levels were within normal ranges and X-ray of the chest were normal. Alpha-fetoprotein and other tumor markers were within normal ranges. At laparoscopy, a strict structure believed to be uterus was noted. The right gonad was found 10 x 15 mm in size, solid, fusiform, on the pelvic sidewall. There was no left gonad. The procedure was terminated following right gonodectomy in June 1999. The patient was transferred to our clinic after the pathology had reported the result as seminoma developed in the atrophic testis.

### RADIATION THERAPY

The patient underwent simulation before treatment. The treatment volume involved bilateral pelvic and paraaortic lymph nodes. The patient treated with an AP-PA technique for a total dose of 26 Gy in 2 Gy daily fractions by using 6 MV photons of linear accelerator.

No recurrence has been found during the postoperative follow-up period of 7 years.

### DISCUSSION

Testicular feminization is a disorder also known as androgen insensitivity syndrome. The 46 XY genetic male totally lacks androgen responsiveness in the target organs thus exhibits a female phenotype. The affected patients have abdominal and inguinal testes, female external genitalia and breasts, blind vaginas, and absent or rudimentary müllerian structures (i.e., fallopian tubes, uterus, and cervix) and the presence of a short vagina.

Testicular feminization results from an androgen resistance involve either the 5 $\alpha$ -reductase enzyme or the androgen receptor. Testicular feminization is usually diagnosed at puberty because of an inguinal hernia or after puberty due to primary amenorrhea. The risk of malignant transformation of the dysgenetic male gonad increases after puberty. The frequency of malignant tumor in testicular feminization has been described to be 5-10% for all ages but increase to 30% by the age of 50. Seminoma followed by embryonal cell carcinoma is the most frequent malignant tumor encountered in undescended testes. Incidental seminoma has also been reported by Sahai et al; in a patient with testicular feminization who underwent bilateral orchiectomy. For this reason, preventive extraction of testis is necessary to prevent malignant transformation in testicular feminization.

### CONCLUSION

Patients with testicular feminization syndrome are frequently affected by benign or malignant tumors in the undescended testes. Surgical removal of the gonads is mandatory to avoid malignant degeneration.

In this report, we presented a case of seminoma developed in a patient with testicular feminization. Diagnosis of testicular feminization with female phenotype and primary amenorrhea was delayed and the patient failed to receive adequate care, resulting in the development of a seminoma.

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