Available online at www.sciencedirect.com

ScienceDirect

journal homepage: www.elsevier.com/locate/apme

Case Report

46, XY complete gonadal dysgenesis (Swyer syndrome): Report of two different cases

Ahmed Samy Elagwany *, *Sally Eltawab*, *Ziad S. Abouzaid*

Department of Obstetrics and Gynecology, Alexandria University, Egypt

ARTICLE INFO

Article history:
Received 4 August 2013
Accepted 7 October 2013
Available online 26 October 2013

Keywords:
Swyer syndrome
Gonadal dysgenesis
Amenorrhea

ABSTRACT

Design: Case report.
Setting: Department of Obstetrics and Gynecology, Alexandria University, Egypt.
Introduction: Swyer's syndrome is a distinct type of pure gonadal dysgenesis characterized by a 46 XY karyotype in female phenotypic patients. It shows an abnormality in testicular differentiation.
Objective: To present cases of Swyer syndrome.
Material and methods: We present the clinical, sonographic, endocrine findings, genetic analyses and treatment in two cases of phenotypic females with XY karyotype and gonadal dysgenesis.
Results: All patients presented with primary amenorrhea. All patients had female-type external genitalia. Secondary sexual characters were developed in all cases. FSH levels were high. Chromosome analyses revealed a 46, XY male karyotype with no detectable mosaicism. The surgical findings were steak gonads, one of them with bilateral gonadoblastoma.
Intervention(s): Bilateral gonadectomy followed by hormone replacement therapy.
Conclusion: We aimed to underline the necessity of considering 46, XY complete pure gonadal dysgenesis in the differential diagnosis in every adolescent female patient with delayed puberty and the importance of early gonadectomy in order to avoid the risk for gonadal tumor development.

1. Introduction

Constitutional causes constitute the most frequent reason for delayed puberty in males while gonadal differentiation disorders or organic diseases are the major causes in females. The lack of appearance of secondary sex characteristics in females and males by the age of 13 and 14, respectively, is described as delayed puberty.[1]

In 1955, Swyer first described two phenotypic women with gonadal dysgenesis without the stigma of Turner syndrome (46 XY pure gonadal dysgenesis, now known as Swyer syndrome).[1,2] The chance of tumor development in Swyer syndrome is 20–30%. The most common tumor described is bilateral gonadoblastoma, but also seen are dysgerminoma and even embryonal carcinoma.[2,3] Five percent of dysgerminomas are discovered in patients who are phenotypically females with abnormal gonads and 46 XY karyotye.[1,3]

* *Corresponding author*. El-Shatby Maternity Hospital, Alexandria University, Alexandria, Egypt. Tel.: +20 1228254247.
E-mail address: Ahmedsamyagwany@gmail.com (A.S. Elagwany).

http://dx.doi.org/10.1016/j.apme.2013.10.005
0976-0016/

In this case report, we aimed to present a case with pure gonadal dysgenesis who presented with complaints of primary amenorrhea and was detected to have bilateral dysgenetic gonads, 46, XY karyotype, as a rare cause of male pseudo-hermaphroditism.

2. Case reports

2.1. *Case 1*

A 16 years old girl was brought to the outpatient department by her parents, due to amenorrhea and poor breast development. The patient did not give any history of cyclical abdominal pain, hormonal intake, radiation exposure, chemotherapy or any central nervous symptoms such as headache or visual disturbances. She gave no history of significant trauma or of having undergone any surgical procedure. There was no history of childhood tuberculosis.

She was the second child of a non-consanguineous marriage and mother's age at the time of delivery was 20 years. On general examination she was 160 cm tall and weighed 65 kg. There was no evidence of acanthosis nigricans, acne, hirsutism, goiter, cushingoid features or Turners stigmata. Her school performance was good. There was no other case with a history of delayed puberty in the family. The remaining systemic examination showed no pathological finding.

Patient had development of breast from 12 years of age. She developed pubic hair from the age of 11 years. Pubic hair was present though the axillary hair was sparse. Her phenotype was completely female including a vaginal opening and there was no evidence of clitoromegaly, the hymen was intact. Examination of secondary sexual characteristics revealed that the breast was in Tanner's Stage 3 (Fig 1b, c).

Ultrasonography (USG) of the pelvis revealed a small sized uterus, no endometrial interface and bilaterally small sized gonadal masses with no follicles, both kidneys are normal. At the time of laparoscopy at a later date revealed both gonadal masses were small hypoplastic uterus and fallopian tubes and examination under anesthesia showed that, the vagina and the cervix were poorly developed.

Routine urine analysis, biochemical parameters, and complete blood count were in normal ranges. The results of endocrinological evaluation were as follows: FSH 37 mIU/mL (N: 2.6–11), LH 17 IU/mL (N: 0.4–7.0) estradiol 10 pg/mL, total testosterone 27 ng/dL (N: 220–800). Her bone age was consistent with the age of 16. A karyotype repeated from two different laboratories showed 46, XY (Fig 1a).

Patient was taken up for an operative laparoscopic procedure under general anesthesia. Intraoperative, the patient had no peritoneal or omental nodule. She also had no deposits over bowel, mesentery and liver surface. And a bilateral gonadectomy was performed (Fig 1d).

Histopathological examination of the both gonadal masses revealed gonadoblastoma. Both fallopian tubes showed

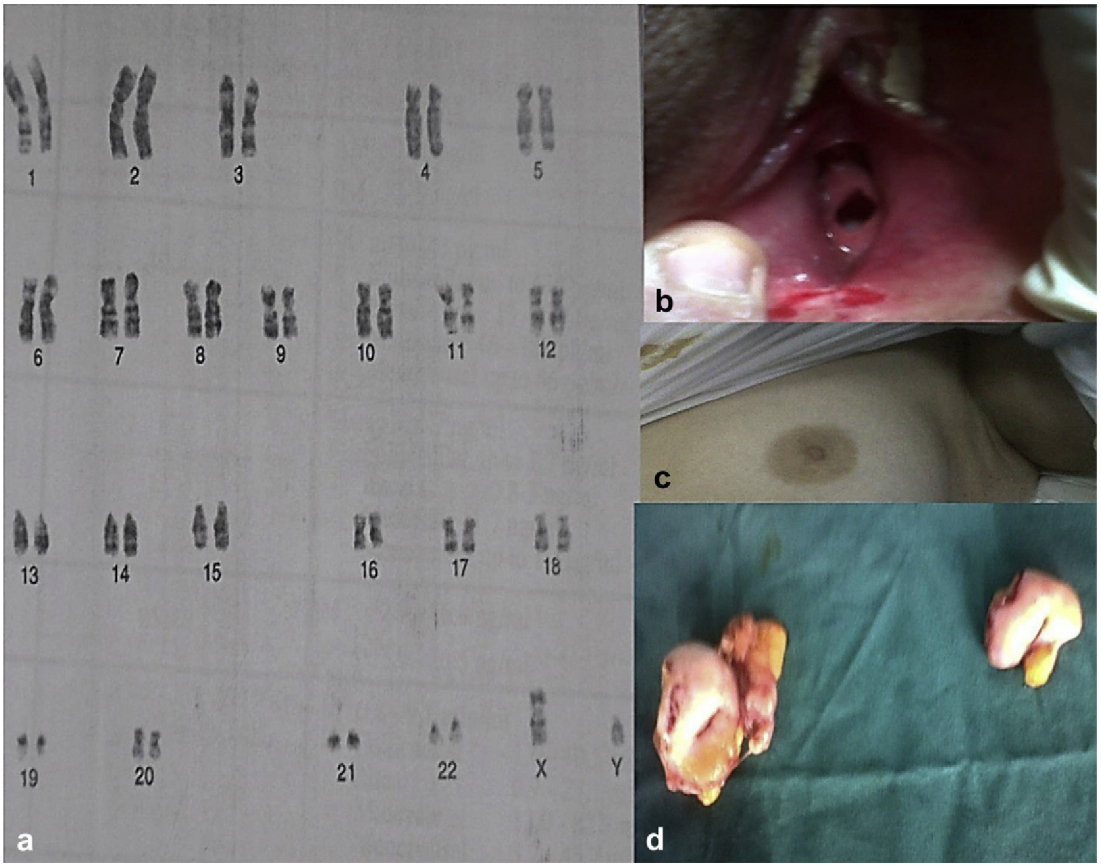

Fig. 1 – 46, XY karyotyping (a), normal external genitalia (b), normal breast (c) and both gonads after resection (d).