Introduction
(Background and Purpose/objectives)

• Disorders of sexual differentiation can be classified as disorders of sexual development and gonadal differentiation (gonadal dysgenesis).
• Gonadal dysgenesis can be complete or partial (Zhu et al., 2011, Nicollo et al., 2005).
• Swyer’s syndrome which is complete gonadal dysgenesis is a sex reversal disorder in which phenotypic females have 46XY karyotype. This was first described by Swyer in 1955 (Swyer 1955).
• Familial cases has rarely been described in the literature (Kempe et al. 2002, Behtash and Karimi Zarchi 2007, Swyer 1955).
• Early diagnosis is of crucial importance given the risk of transformation to malignant germ cell tumour usually dysgerminoma or gonadoblastoma (Zhu et al., 2011).
• It usually becomes apparent during adolescent years (Guidozzi, Ball and Spurdle 1994).

Abstract

We report a case of dysgerminoma diagnosed in a 17-year-old phenotypically female patient with Swyer’s syndrome.

She had complete 46, XY gonadal dysgenesis syndrome.

She had normal McKlerian structures and absent testicular tissue.

She had primary amenorrhea, but matured Tanner stage 5 for breast and pubic hair development.

Urine size was prepubertal.

Description of intervention/study

CASE PRESENTATION

• 17 year old female admitted through ED for complaints of right flank pain x 4 days and fever x 3 days.

• Patient was amenorrheic. Thelarche was at 11 years of age. She has full breast and pubic hair development.

• Family history revealed her 26-year-old sister was also amenorrheic and previously diagnosed with ovarian cancer at age 18.

EXAMINATION FINDINGS

• She was a healthy looking female with eunuchoid habitus.

• Anthropometric measurements were height 158.20cm (+95%), WH 84kg (+95%), BM +25.3.

• Abdominal exam revealed mild tenderness at right lower quadrant.

• She had an abdominal CT scan , CBC, CMP, LH, FSH, TSH, Testosterone level, Estradiol, Prolactin, CEA, CA-15.3, CA-125, CA-19.9 and AFP done.

Chromosomal analysis was also requested.

CT Scan of ovarian mass

Results

• Elevated gonadotropins, low estradiol and normal testosterone levels (Hypergonadotropic hypogonadism).
• CT Scan which showed 11 cm solid partially necrotic mass containing calcification in the cul-de-sac and 6.2cm inflammatory retroperitoneal mass.
• Tumor cells for cytogenetic studies revealed XY male karyotype.
• Chromosomal analysis for karyotyping done showed male karyotype (XY).
• Patient had exploratory laparotomy and the mass was resected and sent for pathology.
• Pathology results showed
  • Enlarged right ovarian tumor weighing 360 grams and measuring approximately 11.0 x 9.5 x 6.5 cm.

Histology and immunohistochemistry

• Cells were positive for PLAP (placental alkaline phosphatase) and CD117.
• It was negative for alpha fetoprotein, CD 30 and HCG.
• The immunoprofile was that of a dysgerminoma without any evidence of a concomitant yolk sac tumor, choriocarcinoma or embryonal carcinoma.

Macroscopically

• Cells had typical features of dysgerminoma.
• Gross
  • CBC
  • CMP
  • TSH, Prolactin, B-HCG, Alpha-fetoprotein
  • CEA, CA-15.3, CA-19.9 and CA125 were all within normal limits.

Histology:

Treatment

Right salpingo-oophorectomy was done. She is presently undergoing chemotherapy with Cyclophosphamide and Bleomycin.

• Hormonal replacement is yet to be done.

Conclusions

• Early diagnosis is of crucial importance given the increased risk of transformation to malignant germ cell cancer usually dysgerminoma or gonadoblastoma.
• High index of suspicion is required for diagnosis.
• Prophylactic bilateral gonadectomy is recommended due to the risk of malignancy; but our patient declined because of future procreation.

References


2012 Texas Pediatric Society Electronic Poster Contest

SWYER’S SYNDROME AND SEQUELAE

Telema Nga MD, Akalolu Amaka MD, Aye Moe MD, Lacaze Mary MD
Department of Pediatrics, Texas Tech University Health Science Centre, Paul L. Foster school of Medicine, El Paso, Texas