True hermaphroditism presenting as bilateral gynecomastia in an adolescent phenotypic male

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Objective: To report a rare case of true hermaphroditism presenting in adolescence as bilateral gynecomastia in a phenotypic male.

Design: Case report.

Setting: Academic medical center.

Patient(s): A 19-year-old phenotypic male who presented with pronounced bilateral gynecomastia and was ultimately diagnosed with true hermaphroditism.

Intervention(s): Endoscopic removal of the internal female genital tract (hemiuterus and ovary), reduction mammoplasty, repair of penile chordee, and left testicular prosthesis placement.

Main Outcome Measure(s): Diagnosis and treatment of true hermaphroditism.

Result(s): Bilateral mammoplasties, repair of a penile chordee, placement of a left testicular prosthesis, and excision of the left hemiuterus and ovary resulted in return to testicular function in this true hermaphrodite.

Conclusion(s): Although rare, true hermaphroditism should be suspected in male patients presenting with bilateral breast enlargement in adolescence. (Fertil Steril 2005;83:1041.e11–e13. ©2005 by American Society for Reproductive Medicine.)

Key Words: True hermaphroditism, gynecomastia, karyotype, chordee, gonadectomy, removal of left hemiuterus

True hermaphroditism is a rare disorder that typically presents as ambiguous genitalia. A true hermaphrodite has both ovarian and testicular tissue present in either the same or the contralateral gonad (1). The karyotype is predominantly 46,XX, and testes, ovaries, and ovotestis can be present in various combinations. As a result, gonads and subsequent reproductive ducts are often asymmetrical (2, 3). The patient’s phenotype dictates the sex of rearing, and this typically depends on which gonad is the dominant one.

The case presented here, however, demonstrates that true hermaphroditism may present later in life not with ambiguous genitalia, but with severe bilateral gynecomastia. In this case, the phenotype and sex of rearing (male) differed from both genetic sex (female) and the dominant gonad (female). We described how surgical correction was accomplished and how hormonal parameters changed following surgery. Institutional review board approval was obtained, and none of the authors had any conflicts of interest.

CASE REPORT

A 19-year-old African male sought medical care for worsening bilateral gynecomastia. He had first experienced bilateral breast bud development at 12 years of age. His gynecomastia progressively worsened over the next 5 years. The patient reported being significantly affected by his gynecomastia and had been socially and intimately inhibited as a consequence. He was highly motivated to receive surgical correction.

At physical examination, the patient was found to be 161.5 cm tall, weighing 128 lb (58 kg). Skin examination showed light facial hair growth. The chest examination revealed severe bilateral gynecomastia (Fig. 1). Both areolas were dilated to 44 millimeters (normal in males: 28 mm [4]). The sternal notch to nipple distance measured 23 cm (normal in males: 19 cm [4]). The inframammary crease to nipple distance was 9 cm. Abundant parenchymal tissue extending toward the outer quadrant was also present. Examination of the genitalia revealed that the phallus was of a normal size, but a chordee was present. Pubic hair was normal. The meatus was patent and normal in appearance. The scrotum was hypoplastic on the left, and no gonad was palpable on that side. The right hemiscrotum was noted to be small but contained a 2 × 2 × 1 cm, smooth gonad. An epididymis-like structure, a spermatic-cord–like structure, and a possible atretic vas were palpable on the right side. No prostate, cervix, or uterus were palpable on examination.

Initial laboratory evaluation revealed a serum follicle-stimulating hormone (FSH) level of 4.6 mIU/mL (normal male level: 1.5 to 12.4) and LH level of 1.2 mIU/mL (normal male level: 1.7 to 8.6). The serum testosterone level was 0.41 ng/mL.
The estradiol level was 51 pg/mL (normal male level: 0 to 45). Chromosome analysis in peripheral blood revealed a 46,XX karyotype; 21 cells were counted, 11 analyzed, and 6 karyotyped. This was followed by a metaphase FISH using a Vysis probe kit for the SRY gene. Fifteen metaphases and 50 interphases were evaluated. The karyotype was 46,XX.ish X cen(CEPx2),Y(SRY-).

Abdominal and pelvic ultrasound demonstrated a 15 × 28 × 37 mm structure posterior to the bladder, reminiscent of a prepubertal uterus with hydrocolpos. A 25 × 29 × 24 mm left ovary with a single dominant follicle measuring 22 × 27 × 23 mm was visualized. A scrotal ultrasound demonstrated a single testis measuring 17 × 19 × 24 mm. The epididymal head measured 6 mm in maximum cephalocaudal dimension.

Magnetic resonance imaging (MRI) of the pelvis confirmed the presence of a small uterus, a normal left ovary, and a single right testis. A prostate was not visualized. Bone age showed all epiphyses to be closed; the bones appeared osteopenic, and the fourth metacarpal was slightly larger than the third.

Bilateral reduction mammoplasty was performed, and approximately 265 grams of tissue were removed from each breast. The surgery was complicated by a minor wound dehiscence of the left breast that required surgical revision under local anesthesia 1 month later.

Right scrotal exploration and right gonadal biopsy to rule out an ovotestis was performed simultaneously with the
reduction mammoplasty. Gonadal biopsies showed parenchyma exclusively testicular in nature. The seminiferous tubules showed exclusively Sertoli cells. A few were infantile in appearance. Leydig cells were noted in the interstitium. No intratubular germ cell neoplasia was identified.

One month later, the patient underwent laparoscopy. The ovary, fallopian tube, and uterus were retroperitoneal. After opening the retroperitoneum and identifying the ureter, left salpingoophorectomy and hysterectomy were performed. The patient also underwent a penile straightening procedure, penoplasty, and insertion of a left testicular prosthesis. Surgical pathology showed an ovary and fallopian tube with no histopathologic abnormality and a small uterus with atrophic endometrium.

Repeat blood results at 6 months after surgery showed elevated gonadotropins with a FSH of 38.7 mIU/mL and LH of 26.6 mIU/mL. Testosterone levels had also increased compared with preoperative levels, now measuring 4.11 ng/mL. The FSH and LH levels remained elevated 1 year later at 30.3 mIU/mL and 16.2 mIU/mL, respectively, despite normal testosterone levels at 3.9 ng/mL. The patient was doing well and no longer had social inhibitions related to his physical appearance.

**DISCUSSION**

True hermaphroditism is rare in North America, but is more common in Africa where this patient was born. Typically, the mode of presentation is one of ambiguous genitalia; the dominant gonad (the ovary) could be safely performed endoscopically after an appropriate radiologic evaluation.

This case illustrates that true hermaphroditism may present in adolescence with bilateral gynecomastia in a patient with a differentiated and functioning gonad of each sex. Moreover, the phenotype and sex of rearing may differ from both the genetic sex and the dominant gonad. Reduction mammoplasty, repair of the chordee, and placement of the testicular prosthesis resulted in a normal male phenotype with adequate testosterone levels despite persistently elevated gonadotropin levels. This case also demonstrates that removal of the internal female genital tract (hemiueterus and ovary) could be safely performed endoscopically after an appropriate radiologic evaluation.

**REFERENCES**