DIAGNOSIS AND MANAGEMENT OF GONADAL MASSES IN YOUNG ADULTS WITH COMPLETE ANDROGEN INSENSITIVITY SYNDROME: A CASE SERIES AND LITERATURE REVIEW

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BACKGROUND

- Early diagnosis and management of gonadal tumors in patients with complete androgen insensitivity syndrome (CAIS) are crucial to improving outcomes
- Clinical exposure to this condition for OBGYNs in training varies depending on the practice setting of their residency program

OBJECTIVE

- **To describe two cases of patients with CAIS complicated by unilateral gonadal masses with delayed management**

MATERIALS & METHODS

- Case series of two CAIS patients, each diagnosed with unilateral gonadal masses at outside hospitals and referred to a safety-net, urban hospital's Reproductive Endocrinology and Infertility (REI) clinic for hormone replacement therapy without a management plan for identified gonadal masses
- A literature review was performed to contextualize the details

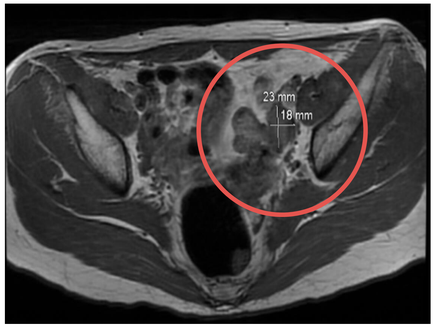
CONCLUSIONS

- **We describe two cases of CAIS resulting in early adulthood gonadal masses with delayed management after discovery**
- Per chart review, neither patient was counseled on the need for gonadectomy before referred to REI clinic
- **Further research is needed to determine evidence-based guidelines on timing of gonadectomy, hormone replacement therapy, and appropriate age-based surveillance in patients with CAIS**
- OBGYNs may benefit from additional training in comprehensive care for CAIS to ensure prompt diagnosis and referral for potential gonadal masses in this population

CASES

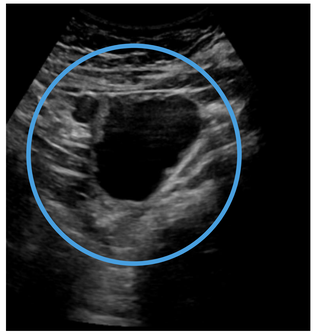
Patient A

- **25-year-old G0** diagnosed with CAIS in the setting of **primary amenorrhea**
- **MRI pelvis** showed an **absent uterus** and a "**2-3 cm left ovarian soft tissue nodule** concerning for a **germ cell tumor**"
- **REI referral** by outside provider for hormone replacement therapy
- **Pelvic mass** noted **on chart review** by REI team
- REI referred patient to urologic oncology for **bilateral gonadectomy**
- Pathology revealed **stage IA seminoma** with no adjuvant treatment required given early stage
- Patient recovered from surgery and started **hormone replacement therapy** as initially desired



Patient B

- **23-year-old G0** with a **family history of CAIS** who was **diagnosed** prenatally on **amniocentesis** and **confirmed at birth**
- Pelvic ultrasound performed in late adolescence revealed a "**3cm left ovarian cyst**" which grew to 5cm and developed a "**septate appearance**" over a 6-year period
- **REI referral** by outside provider for hormone replacement therapy
- Patient **missed scheduled REI appointment**
- REI provider discussed over the phone the importance of prompt evaluation of her gonadal mass
- The patient was **lost to follow up** despite multiple attempts at follow up



LITERATURE REVIEW

- CAIS is an X-linked recessive genetic condition caused by a mutation in the gene that encodes the androgen receptor
- Diagnosis of CAIS relies on both clinical and laboratory data including external female genitalia, primary amenorrhea, absent uterus, undescended testes, 46 XY karyotype, and male-range testosterone levels[1]

Risk of gonadal tumors in CAIS is 2-5% and management includes bilateral gonadectomy with staging[2]

Historically, gonadectomy in CAIS patients is delayed until after puberty and hormone replacement therapy begin after gonadectomy[3]

Studies show that there is a lack of reliable screening modalities for detecting early malignant transformation prior to gonadectomy, thus there are no concrete recommendations on frequency or modality of screening for pre-pubertal, pubertal or young adult CAIS patients[4]

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