Patients with suspected 46,XY CGD based on history and physical exam

- Genetic testing
  - Chromosomal analysis, including karyotype and array CGH
    - FISH for SRY
      - If SRY is present, and chromosome analysis and array CGH are normal, consider sending sequence analysis for SRY, NR5A1, and DHH.
      - If these sequences are normal, verify coverage of NROB1 and WNT4 on array CGH testing and if not well covered, targeted duplication analysis may be considered.
      - If sequences above are normal, consider testing for rare causes including 9p24.3, CBX2 (17q25), and MAP3K1 (5q11.2).

- Hormonal Evaluation
  - Basal LH and FSH may be elevated depending on pubertal status

- Imaging
  - Pelvic Ultrasound or MRI
    - Evaluate for presence and type of Müllerian structures and location of gonads
      - Will show presence of uterus and may show streak gonads

Confirmed CGD

Surgical Management

Proceed to Gonadectomy

Consider sending tumor markers including AFP, LDH, and βhCG if gonadal mass is seen on pre-operative imaging and/or patient presents with discordant pubertal characteristics

Gonadal pathology will reveal a spectrum of under-developed gonads.

---

1. pre-pubertal aged children will likely have LH and FSH that are low (or in the normal range for a pre-pubertal child), whereas post-pubertal aged children will have elevated LH and FSH.

2. The presence of positive tumor markers indicates the necessity for a staged surgical procedure (laparotomy instead of laparoscopy).