Diagnostic Algorithm and Management for Patients with Confirmed XY Gonadal Dysgenesis

History and Physical Exam

Female Phenotype
- Karyotype and Imaging
  - 46,XY Swyer syndrome
    - Recommend Gonadectomy within an ethical framework
  - 46,X/46,XY and variants including mosaic Turner syndrome

Ambiguous Phenotype
- Imaging/Exam/Hormonal Evaluation including hCG stim testing
  - Female Sex Assignment
  - Male Sex Assignment
  - Normal testes on exam (position/palpation) AND normal radiographic appearance?*
    - Yes
    - Orchidopexy with Biopsy if testes are not located in the scrotum
      - LOW Threshold for Gonadectomy
    - No
      - OCT 3/4 + cells on basal lamina, presence of UGT, TSPY strongly +, or SCF +?
        - Yes
          - Recommend Gonadectomy
        - No
          - Routine Yearly Follow-up
            - Consider yearly ultrasound starting at the onset of puberty
            - Consider one post-pubertal biopsy with immunohistochemical staining.

Mildly Undervirilized Male Phenotype
- Imaging/Exam
  - Normal testes on exam (position/palpation) AND normal radiographic appearance?*
    - Yes
    - Routine Yearly Physical Exam and regular self-exam
    - No
    - Orchidopexy with Biopsy

Fully Virilized Male Phenotype
- Imaging/Exam
  - Normal testes on exam (position/palpation) AND normal radiographic appearance?*
    - Yes
    - Routine Yearly Physical Exam and regular self-exam
    - No
    - Orchidopexy with Biopsy

* Applies to each gonad. In cases where one gonad is dysgenetic and the other appears normal, recommend removal of the dysgenetic testes and routine monitoring of the normal-appearing testes