Male Pseudohermaphroditism

A 15-year-old female patient with primary amenorrhea and absent secondary sexual characters.

**History:** A 15-year-old female patient with primary amenorrhea and absent secondary sexual characters.
Figure 1: Sagittal T2WI shows completely absent uterus or vagina (arrows)
Figure 2: Coronal T2WI shows feminine external genitalia (arrows)

Figure 3: Axial T2WI shows feminine perineum with absent male external genitalia
Figure 4: Coronal T2WI and post contrast T1 fat sat show bilateral inguinal testis (arrows)

**Discussion:** Disorders of sex development (DSDs) are congenital conditions in which the
development of chromosomal, gonadal, or anatomic sex is atypical. DSDs can be classified broadly
into four categories on the basis of gonadal histologic features:
(a) female pseudohermaphroditism (46,XX with two ovaries);
(b) male pseudohermaphroditism (46,XY with two testes);
(c) true hermaphroditism (ovotesticular DSD) (both ovarian and testicular tissues); and
(d) gonadal dysgenesis, either mixed (a testis and a streak gonad) or pure (bilateral streak gonads).
Current methods for diagnosing DSD include a thorough physical examination, laboratory tests to
determine hormone levels and identify chromosomal abnormalities, and radiologic imaging of the
genitourinary tract and adjacent organs.

Ultrasonography is the primary modality for demonstrating internal organs; Genitography is used to
assess the urethra, vagina, and any fistulas or complex tracts; Magnetic resonance imaging is used
as an adjunct modality to assess for internal gonads and genitalia. Early and appropriate gender
assignment is necessary for healthy physical and psychologic development of children with
ambiguous genitalia.

**Classification and New Terminology:**
A new nomenclature and classification system as well as
new management recommendations for DSD was introduced. These disorders were further
subdivided into 46,XY DSD (disorders of gonadal or testicular development and impaired androgen
synthesis or action), 46,XX DSD (disorders of gonadal or ovarian development and androgen excess),
and chromosomal DSD (numeric sex chromosome anomalies). There is some overlap between these
three subgroups. This new terminology has replaced the older terms hermaphroditism and
pseudohermaphroditism and emphasizes the genetic origin of the disorders.

**Diagnosis:** Old terminology: Male pseudohermaphroditism, XY male undermasculinization; New
terminology: 46,XY DSD

**Finding:** Male gonadal development is abnormal, androgen synthesis or action is deficient, external
genitalia are undermasculinized to a variable degree.

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