Male Pseudohermaphroditism

By Tougan Taha, MD [2]

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
**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.

Tougan Taha MD
*Lecturer of Radiodiagnosis*
*Ain Shams University*
*Cairo, Egypt*

**Source URL:** [http://www.diagnosticimaging.com/case-studies/male-pseudohermaphroditism](http://www.diagnosticimaging.com/case-studies/male-pseudohermaphroditism)

**Links:**
[1] [http://www.diagnosticimaging.com/case-studies](http://www.diagnosticimaging.com/case-studies)
[2] [http://www.diagnosticimaging.com/authors/tougan-taha-md](http://www.diagnosticimaging.com/authors/tougan-taha-md)