Testicular feminization - Complete androgen insensitivity syndrome

19-year-old female patient presented because of primary amenorrhea with absent pubic hair. She had normal breasts with scanty axillary hair.

Imaging Findings:

Ultrasonography elsewhere suggested absent uterus and ovaries. Multi-echo multi-planar MRI of the pelvis was performed on 3 Tesla MR scanner, using 6 channel SENSE torso coil. The findings were as follows:

- Absence of uterus and ovaries in the pelvis
- Vagina was seen in normal position in between urinary bladder and rectum
- Presence of undescended testes in both the inguinal canals

Discussion:

Background: Testicular feminization, or the Androgen Insensitivity Syndrome (AIS), is a X-linked recessive androgen receptor (AR) disorder, when a man, genetically XY, has some physical characteristics of a woman, or even a full female phenotype. These individuals have absence of Mullerian duct derivatives i.e. uterus, fallopian tubes, cervix and upper vagina with presence of intraabdominal, inguinal and labial testes [1].

The presence of SRY gene on Y chromosome directs the gonad to become a testicle, and produce the Sertoli and Leydig cells. Due to secretion of Mullerian Inhibiting Factor (MIF) by testicular sertoli cells, they do not develop Mullerian structures. Leydig cells secrete testosterone and would stabilize the Wolffian ducts and cause masculinization of the external genitalia, however, in AIS, there is AR gene mutation resulting in defecting binding of testosterone and lack of masculinization of external genitalia [2].

Clinical Features: Depending on the type of AR mutation, the failure of sexual differentiation can be either complete (CAIS), partial (PAIS), or mild (MAIS). CAIS individuals have female phenotype with normal breasts, scanty axillary and pubic hair and present with primary amenorrhoea.

Imaging: Transabdominal ultrasonography is useful is showing absence of Mullerian structures and presence of testes. However, it is operator-dependent and can be inconclusive. MRI plays an important role in confirmation and correct localisation of testes - intra-abdominal, sublabial, inguinal locations.

Karyotyping shows male (XY) pattern in individuals with a female phenotype; confirming the diagnosis of CAIS.

PAIS shows presence of hypospadic micropenis and a bifid scrotum that may contain the testes, whereas MIAS is not associated with male genital anomalies but presents as infertility [1].

Differential diagnosis: Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome shows normal female phenotype with
female karyotype (XX) and normal ovaries. These patients have congenital absence of uterus, cervix and upper vagina.

Outcome: Since there is risk of malignancy developing in undescended testes, which increases with age, surgery is recommended and puberty is artificially induced with oestrogen supplements.

Take home message: Complete androgen insensitivity syndrome is a rare disorder characterised by male karyotype with female phenotype. This disorder should be kept in mind while evaluating young women for primary amenorrhea, and MRI is the gold standard for its diagnosis.

**Differential Diagnosis List:** Testicular feminization - Complete androgen insensitivity syndrome, Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome, Partial/Mild androgen insensitivity syndrome

**Final Diagnosis:** Testicular feminization - Complete androgen insensitivity syndrome

**References:**

Marco Nezzo, Pieter De Visschere, Guy T'Sjoen, Steven Weyers, and Geert Villeirs (2013) “Role of Imaging in the Diagnosis and Management of Complete Androgen Insensitivity Syndrome in Adults,. Case Reports in Radiology, vol. 2013, Article ID 158484, 6 pages, 2013. (PMID: [23762728](https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3884482/))

Sana Khan and LaTasha B Craig (2013) A Review of Radiologic Imaging in Patients with Androgen Insensitivity. J Genit Syst Disor S1
Description: T1W image - axial view showing presence of undescended testis (blue arrows) in the inguinal canals. Origin: Lifescan Imaging centre, Mumbai, India.
Description: T2W axial view shows presence of vaginal opening (red arrow) between urethral orifices (green arrow) and anus (yellow arrow). Also note the presence of undescended testes (blue arrows) in both inguinal canals. Origin: Lifescan Imaging centre, Mumbai
Description: Note the absence of both the ovaries (circles) on this T2W coronal view. Ileal loops are seen in the pelvis. Origin: Lifescan Imaging centre, Mumbai
Description: T2W coronal view shows presence of undescended testes (blue arrows) in both the inguinal canals. Origin: Lifescan Imaging centre, Mumbai
**Figure 5**

Description: T2W sagittal view showing presence of vagina (red arrow) in normal anatomical location between urinary bladder (green arrow) and rectum (yellow arrow). No uterus is visualised. **Origin:** Lifescan Imaging centre, Mumbai, India