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Usefulness and role of magnetic resonance imaging in a case of complete androgen insensitivity syndrome

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Complete androgen insensitivity syndrome (CAIS) is an X-linked, recessive disorder caused by mutations of the androgen receptor (AR), in which genetic males (46,XY) show female external genitalia. Individuals with CAIS have mostly normal external genitalia, lack of Müllerian structures (Fallopian tubes, uterus, proximal portion of the vagina) and undescended testes (intra-abdominal, inguinal, or labial). Management and diagnosis of CAIS should be undertaken by a multidisciplinary team of experts in sexual development disorders. Gonadectomy represents a standard therapeutic choice to prevent testicular malignancy in the prepubertal period, with subsequent hormonal replacement therapy, or in late adolescence, after completion of pubertal development. Imaging examinations play a pivotal role in the diagnosis, assessment, and detection of the gonads before surgical treatments. Magnetic resonance imaging (MRI) is the gold standard to diagnose and locate the gonads, and to plan laparoscopic gonadectomy and gonadic surveillance, in particular in the increasingly large number of patients who decide to delay or ultimately not to undergo gonadectomy. We present a case of a 14-year-old female with primary amenorrhea.

Case report

A 14-year-old phenotypic female was referred to our hospital with primary amenorrhea and delayed pubertal development. She was 172 cm tall (above the 95th percentile) and weighed 88.5 kg (above the 95th percentile); her BMI was 29.9 kg/m^2 (growth above the genetic target and the normal female population). The patient, born at term by spontaneous delivery, reported repeated episodes of narcolepsy and lipothymia during physical activities and a single episode of plausible vaginal bleeding at 10 years. A physical exam revealed Tanner stage B3P2, with little pubic and axillary hair. The pelvic exam demonstrated hypoplastic labia majora, a blind-ended vagina, and the absence of uterus and cervix. Laboratory tests showed typical hormonal changes for age, sex, and pubertal stage: high testosterone (26.3 ng/dl), DHEAS (205.9 µg/dl), and LH (121 mUI/ml), with low FSH (12 mUI/ml) and normal estradiol (16 pg/ml); the karyotype analysis was 46,XY.

A transabdominal ultrasonography exam of the pelvis, performed in another hospital, demonstrated the absence of the uterus and gonadal structures. MRI (Achieva®, Philips Medical System, Eindhoven, the Netherlands, 1.5 T) was performed with axial T1W, axial and sagittal T2W turbo spin echo HR (high resolution), and coronal STIR TSE and axial diffusion-weighted imaging (DWI) (b values: 0 and 1000 s/mm^2) sequences. Conventional sequences demonstrated two homogeneous ovoid solid masses, hypointense on T1 and iso-hyperintense on T2, in the ingui-

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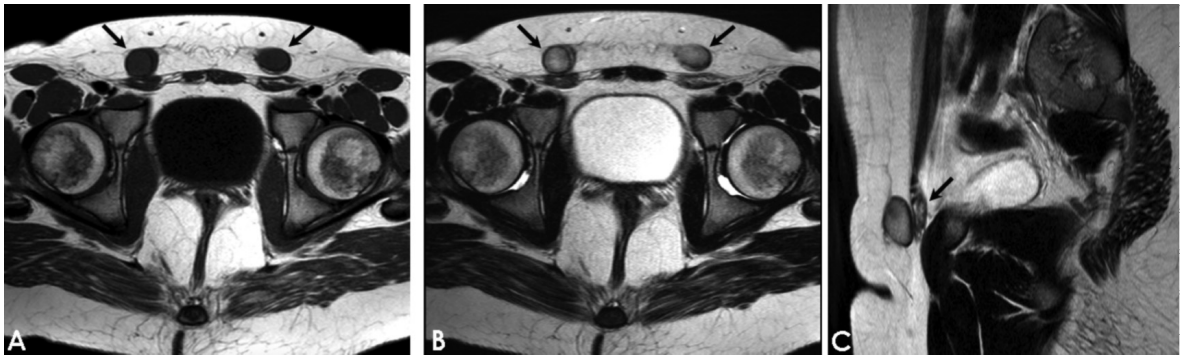

Fig. 1. (A-B) Axial T1- and T2-weighted images reveal bilateral solid nodular structures with homogeneous, mild hyperintensity in the anterior lower pelvic wall, adjacent to the external inguinal ring, compatible with testes (black arrow); (C) Sagittal T2 TSE image shows subtle, hyperintense, tubular structure similar to atrophic epididymides behind the gonads (black arrow).

nal canal close to the external ring, consistent with testes (Fig. 1, A-B). The right gonad measured 1.8x1.9x3.1 cm, and the left measured 1.7x2.4x2.9 cm. Tubular, subtle, T2-hyperintense structures, adjacent to the gonads, were seen; they appeared to be similar in size and shape to atrophic epididymides (Fig. 1C). There was no evidence of the uterus in the pelvis; sagittal T2W TSE demonstrated the presence of a short, blind-ended vagina between the bladder and (posteriorly) the rectum. In the recto-uterine pouch, there was free fluid collection (Fig. 2). DWI (b=1000) showed hyperintense extrapelvic gonads due to restricted diffusion, and low values on the ADC map (0.8 mm^2/s on the right and 0.9 mm^2/s on the left) (Fig. 3). The patient subsequently underwent hormonal replacement therapy and received psychosocial support.

Commonly included in the differential diagnosis are other causes of primary amenorrhea, in particular the Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome, characterized by Müllerian duct anomalies (congenital absence of the upper part of the vagina, uterine agenesis, or

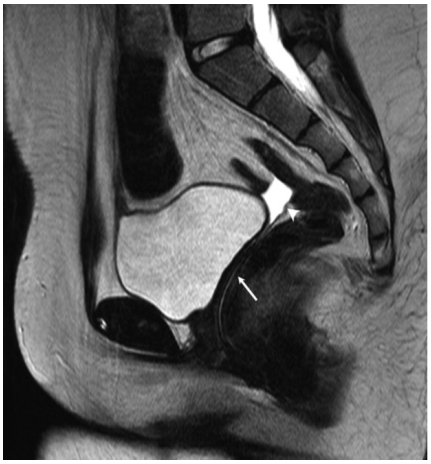

Fig. 2. Sagittal T2 TSE shows a blind-ended vagina (white arrow) between the bladder and the rectum posteriorly and the absence of Müllerian structures, such as the uterus; a fluid collection fills the Douglas pouch (white arrowhead).

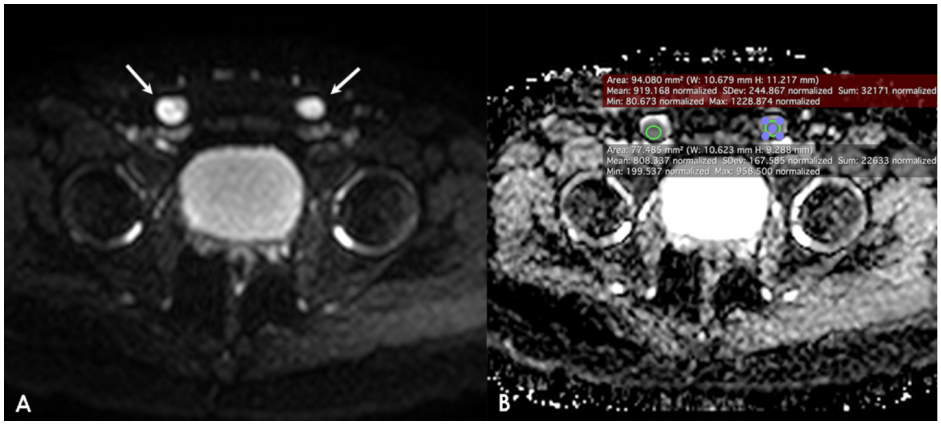

Fig. 3. DWI demonstrates hyperintense signal of the extrapelvic gonads; ADC map shows bilateral low signal intensity with 0.8 mm^2/s on the right and 0.9 mm^2/s on the left. No signs of malignant degeneration are detected.

the presence of a rudimentary uterus), a female karyotype, normal female external genitalia, normal breast development, and normal axillary and pubic hair. In Swyer syndrome (or 46,XY complete gonadal dysgenesis), the patient presents with normal female external genitalia, lack of breast development, and short stature with completely undeveloped ("streak") gonads and the presence of Müllerian structures. Partial androgen insensitivity syndrome