CASE REPORT

Ultrasound in Ambiguous Genitalia

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Received 24 May, 2010; accepted 10 December, 2010

KEY WORDS
ambiguous genitalia, congenital adrenal hyperplasia, Müllerian remnant syndrome, testicular feminization syndrome, ultrasound

Case reports

Case 1

This patient was a 1-month-old infant with a small penis and poorly developed scrotum (Fig. 1). There was also a small sinus tract at the base of the penile shaft. Ultrasound revealed bilateral testes located low in the inguinal region, which revealed multitude echoes and thus appeared abnormal (Fig. 2). There was also the presence of a uterus and cervix behind the bladder. The cervical canal was seen communicating with the sinus tract at the base of the penis. No ovarian tissue was seen (Fig. 3). A diagnosis of persistent Müllerian remnant syndrome was made.

Case 2

A 7-year-old girl was brought to our attention due to coarsening of her facial features, development of pubic hair and enlargement of her clitoris. She also had mild clitoral fusion (Fig. 4). On transabdominal ultrasound, a normal uterus and ovaries were identified. There was no evidence of any pelvic mass. Evaluation of the adrenal bed revealed bilateral enlarged adrenal glands (Fig. 5). This suggested a diagnosis of congenital adrenal hyperplasia. Further endocrine evaluation confirmed 21-hydroxylase deficiency.
Case 3

A thirteen year old girl with normal female genitalia was being investigated for delayed menarche. A transabdominal ultrasound revealed the absence of the uterus and ovaries. A high resolution ultrasound of the labial folds revealed a testicle in both of the labial folds (Fig. 6). A diagnosis of testicular feminization syndrome was made. Genetic workup and hormonal tests helped in confirming this diagnosis.

All the cases were evaluated with a GE LOGIQ5 Expert (Milwaukee, Wisconsin, USA) with curvilinear and high resolution transducers.

Discussion

Ambiguous genitalia is a term for a rare condition in which the newborn external genitalia do not conform to either the male or female type. This results in serious psycho-social concerns, resulting in multitude of complex tests for characterization and management. Because of its availability, ultrasound can be used as the first-line diagnostic tool to help in assigning the sex of the individual.

Mehdi [1] evaluated 12 cases of ambiguous genitalia with ultrasound and magnetic resonance imaging. The laboratory tests and surgical evidence proved that the imaging results were in agreement and concluded that ultrasound is the primary imaging modality for the evaluation of the internal reproductive organs.

Persistent Müllerian remnant syndrome refers to a form of internal male pseudohermaphroditism characterized by...
the presence of Müllerian duct derivatives in an otherwise normal male patient [2].

Our first case revealed the presence of Müllerian derivatives in a genotypic male infant. The condition is caused by an insufficient amount of anti-Müllerian hormone released in the male fetus from the 8th to the 10th week of gestation or by the insensitivity of the target organ for this factor [3].

Persistent Müllerian duct syndrome occurs in two different anatomic forms. The partially descended testicles type (80–90% of reported cases) occurs with unilateral cryptorchidism and contralateral inguinal hernia. One testicle has descended into the scrotum and the ipsilateral uterus and fallopian tube have entered the inguinal canal; a condition known as hernia uteri inguinalis. Sometimes the opposite testicle is dragged by these structures to the same hemiscrotum (transverse testicular ectopia). The undescended testes form, as in our case, into bilateral non-palpable testes located in a high position. The uterus is fixed in the pelvis and both testes are embedded in the round ligament [4].

Our second case, which was diagnosed as congenital adrenal hyperplasia (CAH), was caused by a deficiency of enzymes of the cortisol pathway. The most frequent is steroid 21-hydroxylase deficiency, accounting for more than 90% of cases [5]. It is the most common cause of female pseudohermaphroditism. Although there are few reports which demonstrate enlarged adrenal glands by ultrasound in cases of congenital adrenal hyperplasia, ultrasound can still be used for evaluation of the uterus and the adrenal bed.

Bryan et al [6] evaluated the role of ultrasound in the evaluation of CAH in new born infants. Ultrasonic examinations were performed on eight newborn infants with ambiguous genitalia who were subsequently proven to have adrenogonadal syndrome. The adrenals were found to be enlarged in three patients, at the upper limit of normal in three patients, and in two infants the adrenals were well within normal limits in size.

Al-Alwan et al retrospectively evaluated 52 cases of ambiguous genitalia and concluded that adrenal ultrasonography is a highly sensitive and specific adjunct in the diagnosis of CAH. The presence of enlarged, lobulated adrenal glands with stippled echogenicity is invariably associated with CAH [7].

Our third case was diagnosed as testicular feminization syndrome, which is also called androgen insensitivity syndrome. It is characterized by a female phenotype with normal breast development and female external genitalia.

Fig. 5 Ultrasound shows enlarged adrenal glands.

Fig. 6 Testicles in the labial fold. (A) The right testicle shows in the right labial fold; (B) The left testicle shows in the left labial fold.
but with primary amenorrhea, paucity of pubic and other body hair, a blind vaginal pouch and intra-abdominal or inguinal testes. It occurs due to mutations in the steroid 5-alpha-reductase type 2 gene (SRD5A2), presenting at birth with variable degrees of under-virilization [8].

Testicular feminization syndrome may present as complete form and incomplete form. In the complete form there is no androgen response, therefore normal external female genitalia develop and these infants are raised as female. There can be labial or inguinal swellings which contain testes. These patients most often present in late adolescence with primary amenorrhea. There is absence of the uterus and ovaries on ultrasound or laparoscopy [9].

Ultrasonography is the imaging modality of choice for the evaluation of patients with primary amenorrhea. Careful correlation with clinical history, physical examination and laboratory findings significantly narrows the diagnostic possibilities thus allowing for a more precise diagnosis [10].

Ultrasoundography is also the primary modality for establishing the presence or absence of the gonads and Müllerian derivatives. It can be performed quickly and does not involve radiation. Ultrasound examination should include inguinal, perineal, renal and adrenal regions [11].

In conclusion, ultrasound evaluation should be considered in patients presenting with ambiguous genitalia. It helps in identifying the type of reproductive organs present, thus narrowing the differentials, and in deciding further tests and management.

References