Journal of Medical Ultrasound (2011) **19**, 131—134

Available online at www.sciencedirect.com

SciVerse ScienceDirect

journal homepage: www.jmu-online.com

CASE REPORT

Ultrasound in Ambiguous Genitalia

Muktachand L. Rokade*

Jupiter Hospital, Thane, India

Received 24 May, 2010; accepted 10 December, 2010

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

Ambiguous genitalia is a medical term for rare condition in which the newborn's external genitalia do not conform to either male or female type. The condition of ambiguous genitalia has serious psycho-social concerns and is usually followed-up with a multitude of complex tests for identifying the gender and the cause of the anomaly. Three cases of ambiguous genitalia are reported here where ultrasound helped to elucidate the probable cause and to direct further appropriate tests.

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.

* Dr Muktachand L. Rokade MBBS, MD, DNB, Jupiter Hospital, Eastern Express Highway, Thane 400601, India.
E-mail address: drmlrokade@gmail.com.

0929-6441 © 2011, Elsevier Taiwan LLC and the Chinese Taipei Society of Ultrasound in Medicine. Open access under CC BY-NC-ND license.
doi:10.1016/j.jmu.2011.10.003

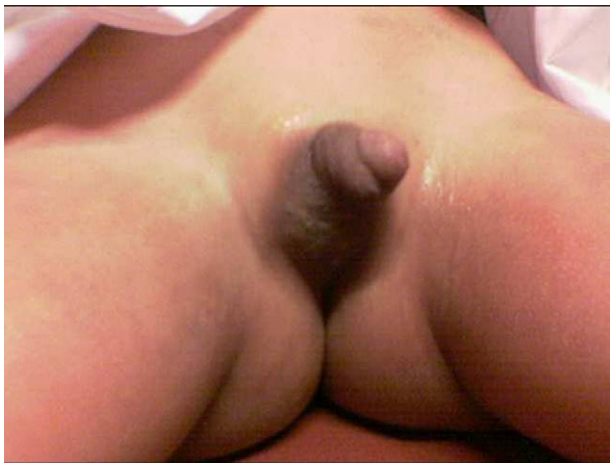

Fig. 1 Clinical photograph shows small penis and poorly developed scrotum.

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.

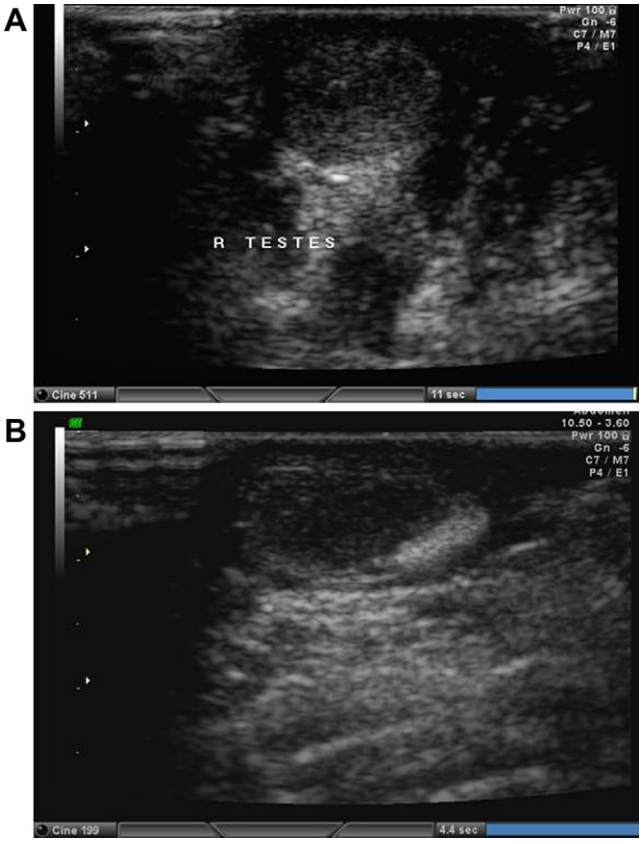

Fig. 2 Ultrasonographic findings of bilateral small testicles with hyperechoic foci. (A) Right testis; (B) left testis.

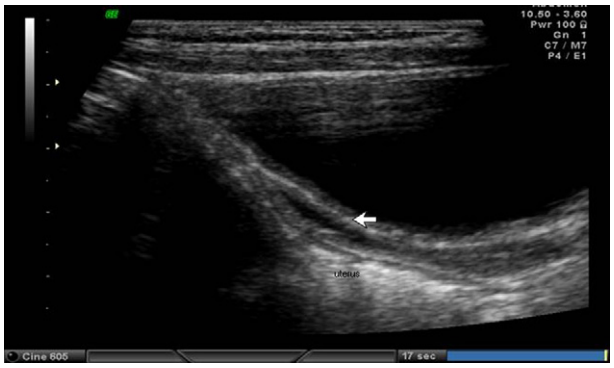

Fig. 3 Presence of uterus and cervix posterior to the bladder. Note the fluid filled cervical canal communicating with the sinus tract.

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

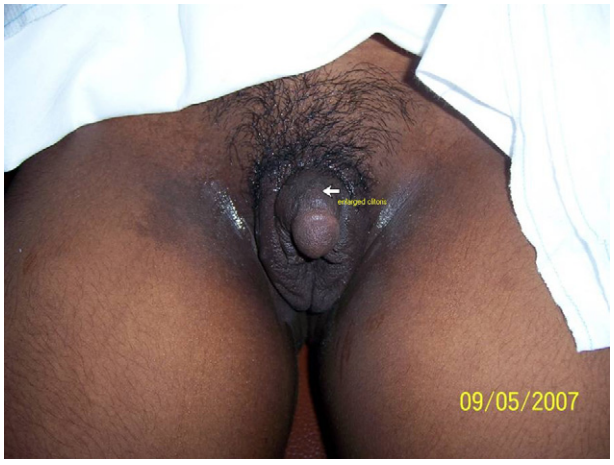

Fig. 4 Clinical photograph showing ambiguous genitalia with enlarged clitoris and labial fusion.