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Ambiguous genitalia: comparative role of pelvic ultrasonography and genitography

N. A. M. AL JURAYYAN, P. J. PATEL*, A. S. AL HERBISH, M. A. ABDULLAH**, A. M. ABO-BAKR, A. A. AL RABEEAH†, A. J. JAWAD† & A. I. AL SAMMARAI†

*Division of Paediatric Endocrinology, *Department of Radiology and †Division of Paediatric Surgery, College of Medicine, King Saud University, and **Department of Paediatrics, Security Forces Hospital, Riyadh, Saudi Arabia*

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Summary Ambiguous genitalia represents a true medical and social emergency which needs a multi-disciplinary team approach for elucidation. The paediatric radiologist plays an important role in defining the genital anatomy which remains one of the most important factors in sex determination. Aiming to compare the predictive value of pelvic ultrasonography and genitography in sex determination in patients with ambiguous genitalia, we retrospectively reviewed the medical records of 69 patients, 53 females and 16 males, where both procedures were employed. In female pseudohermaphroditism, the presence of a uterus with or without vagina was predicted in 46 (86.8%) patients by ultrasound compared with 44 (83%) patients in whom a genitogram revealed a vagina ± uterus. In six (11.3%) patients, a genitogram revealed a male-type urethra. The combination of ultrasound and genitogram, however, was more sensitive and predicted the presence of a uterus with or without vagina in 52 (98.1%) patients. In male pseudohermaphroditism, there was no false positive by ultrasound, and a genitogram revealed a male-type urethra in 12 (75%) patients. In conclusion, although real time pelvic ultrasonography is less invasive than genitography, its yield in elucidating genital anatomy is comparable. The combination of both procedures is more informative and has a better yield.

Introduction

Ambiguous genitalia in a newborn is a medical and social emergency requiring a multi-disciplinary team approach. Not only might there be immediate physiological problems such as shock, hypoglycaemia or subsequent salt loss, but there is also a need to assign a sex. Failure to do so can lead to wrong sex assignment with grave consequences.[1–3] The paediatric radiologist plays a significant role in elucidating the existing anatomy, which, in conjunction with chromosomal analysis, can lead to appropriate sex assignment and, hence, optimal therapy.[1,3–14]

We have reviewed our experience with 69 children with ambiguous genitalia in whom pelvic ultrasonography, genitography and chromosomal and appropriate hormonal investigations were employed. Our aim was to compare the prediction rate of pelvic ultrasonography and genitography and their value in sex determination. To our knowledge, no similar study has been reported in the English literature.

Materials and methods

Eighty-one children with ambiguous genitalia

Reprint requests to: Dr Nasir A. M. Al Jurayyan, Department of Paediatrics (39), College of Medicine, King Saud University, PO Box 2925, Riyadh 11461, Saudi Arabia.

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TABLE I. Aetiological diagnosis and radiological (ultrasound and genitogram) studies in 53 patients with female pseudohermaphroditism (46 XX karyotype)

Aetiological diagnosis (no.)	Ultrasound	Genitogram		
	+ ve uterus	+ ve vagina ± uterus	Male-type urethra	Inc.
CAH-21-hydroxylase deficiency (38)	32	32	4	2
CAH-11-hydroxylase deficiency (12)	11	9	2	1
Isolated clitoral hypertrophy (3)	3	3	—	—
Total (53)	46 (86.8%)	44 (83%)	6 (11.3%)	3 (5.6%)

CAH = Congenital adrenal hyperplasia; Inc. = inconclusive.

were managed between 1984 and 1994 at the Endocrine Unit, Department of Paediatrics, King Khalid University Hospital (KKUH), Riyadh, Saudi Arabia. In 69 cases, both pelvic ultrasonography and genitography were employed and they therefore constituted the subject of this study. In all of them, the aetiological diagnosis was confirmed by chromosomal studies and appropriate hormonal investigations.[2,3] The clinical findings, including the degree of virilization,[15] were correlated with the radiological findings.

Ultrasound examinations were performed using real time sector scanners with either 5 or 7.5 MHz transducers. The aim of the examination was to determine the presence or absence of a uterus. The ovaries were more difficult to identify with certainty than the uterus, and were therefore not relied upon in determining the gender. An attempt was made to identify the uterus in both sagittal and transverse planes in each case. The children were not catheterized in order to fill their bladders, but studies were performed after sufficient urine had been allowed to accumulate.

Genitography is a more invasive procedure involving cannulation of the urethra and urogenital sinus. A preliminary film of the pelvis was obtained in the supine and lateral positions. After sterile cleaning, water-soluble non-ionic contrast medium was injected through a feeding tube into the perineal orifices. When it had filled the vagina, the medium entered the endocervical canal and, in some cases, the uterine cavity, the fallopian tubes and/or the urethra. All examinations were performed under fluoroscopy in the lateral position with a spot film device.

Results

Sixty-nine children satisfied the study criteria. Their ages ranged from a few days to 10 years. There were 53 children with female pseudohermaphroditism (46 XX karyotype) and 16 with male pseudohermaphroditism (46 XY karyotype). Tables I and II show the genetic sex, aetiological diagnosis and radiological (ultrasound and genitogram) appearances.

In the female pseudohermaphroditism group, the uterus was detected by ultrasound in 46 (86.8%) children (Fig. 1), giving a false negative result of 13.2%. The genitogram revealed a vagina with or without a uterus in 44 (83%) patients (Fig. 2); a male-type urethra, with no evidence of uterus or vagina, was observed in six (11.3%); in the other three the study was inconclusive. All were noted to have severe external virilization. In combining the ultrasonography and genitography studies, the yield rate was 98.1%.

In one child, the uterus was not visualized with ultrasound and the genitogram revealed a male-type urethra. This child was initially assigned male sex as a result of a severe degree

TABLE II. Aetiological diagnosis and radiological (ultrasound and genitogram) studies in 16 patients with male pseudohermaphroditism (46 XY karyotype)

Aetiological diagnosis (no.)	Ultrasound	Genitogram		
	− ve uterus	− ve vagina and uterus	Male-type urethra	Inc.
Organ unresponsiveness (8)	8	8	4	4
5 α-reductase deficiency (4)	4	4	4	—
Hypopituitarism (2)	2	2	2	—
Isolated hypospadias (1)	1	1	1	—
CAH 3-β-hydroxysteroid dehydrogenase deficiency (1)	1	1	1	—
Total (16)	16 (100%)	16 (100%)	12 (75%)	4 (25%)

CAH = Congenital adrenal hyperplasia; Inc. = inconclusive.

of virilization. The child was referred to our hospital at 3 months of age, and eventually diagnosed as having female pseudohermaphroditism owing to 21-hydroxylase deficiency congenital adrenal hyperplasia.

In the male pseudohermaphroditism group, ultrasonography revealed no false positive result for a uterus. Genitogram revealed a male-type urethra in only 12 (75%). The study was inconclusive in four (25%). All proved to have the testicular feminization syndrome.

Discussion

Ambiguous genitalia in a neonate is a medical and social emergency, challenging the physician to arrive expediently at a rational sex assignment. Although physical examination remains of great importance in the initial evaluation, it is still difficult to distinguish clearly between male and female pseudohermaphroditism, especially with the wide spectrum of variations in the physical appearances. Some affected male infants develop a female phenotype, with complete failure of virilization of the Wollfian ducts, urogenital sinus and external genitalia. On the other hand, female infants may appear to be entirely normal boys with bilateral undescended testicles.[4–3,15] Furthermore, biochemical and chromosomal confirmation usually takes a few days, and this delay causes increased emotional stress in the parents.[1–3]

Radiological evaluation, unlike other examinations, has proved to be capable of demonstrating the appropriate anatomy—the presence or absence of Müllerian structures,[1,3–14] considered of paramount importance in gender assignment and preparation for surgery. Demonstration of the Müllerian structures is usually achieved by sonography.

If Müllerian structures are present, the individual obviously does not have male pseudohermaphroditism with Müllerian regression; there is either female pseudohermaphroditism, which is the most common, or, rarely, male pseudohermaphroditism without Müllerian regression, or true hermaphroditism. Sonography is very helpful in identifying the presence of the cervix and uterus, which are usually prominent in the early days of life.[1,3,5–7,11,14] However, the absence of the uterus on sonogram or genitogram does not prove that the uterus is not present. It is very important that the bladder is full when the study is done. Genitography usually provides further details of the urogenital sinus and vaginal structures.[4,9,10,12] However, it is not unusual for the Müllerian structures to be demonstrated by one study and not by the other.[1,3] Abdullah *et al.* have shown, in a small series, that combining ultrasonography with a genitogram gave a yield rate for the uterus of almost 100%.[1]

Our results show that the uterus can be identified by ultrasound in a high percentage (87%) of female infants, and none of the

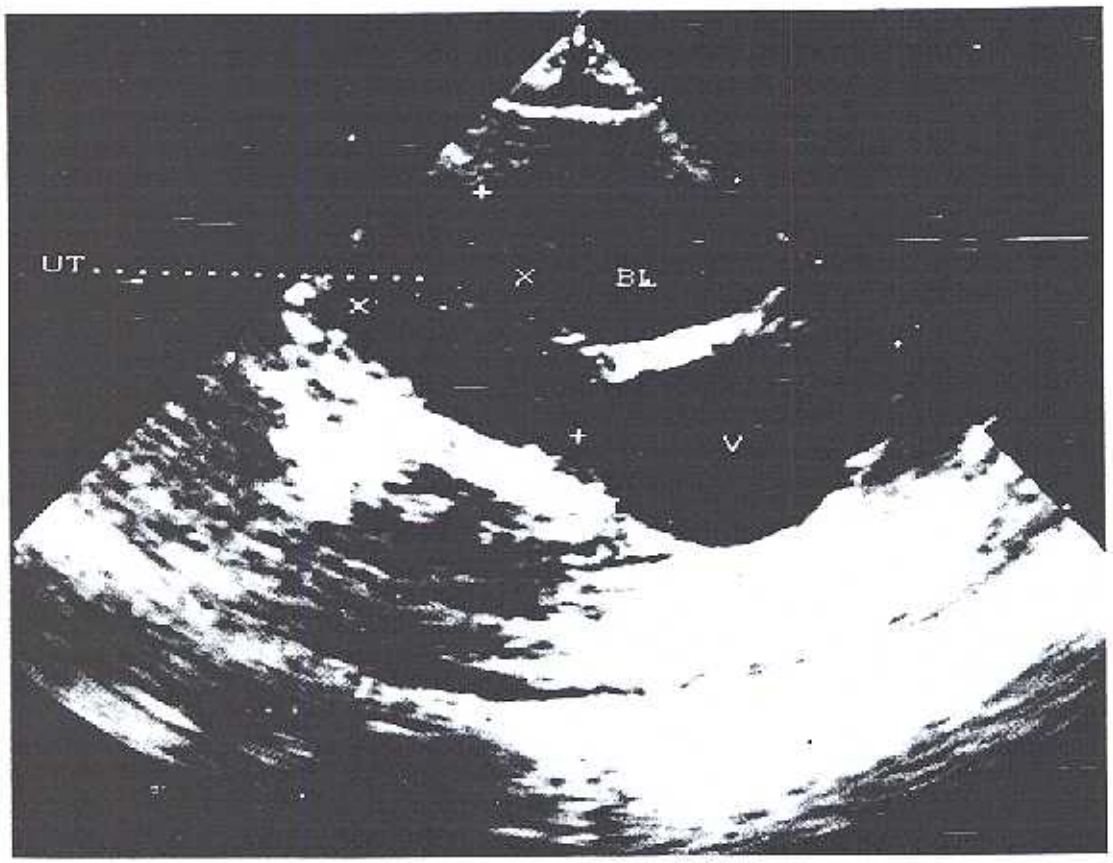

FIG. 1. Ultrasound (sagittal) of pelvis showing anteriorly bladder (BL), postero-superiorly uterus (UT), and postero-inferiorly vagina (V).

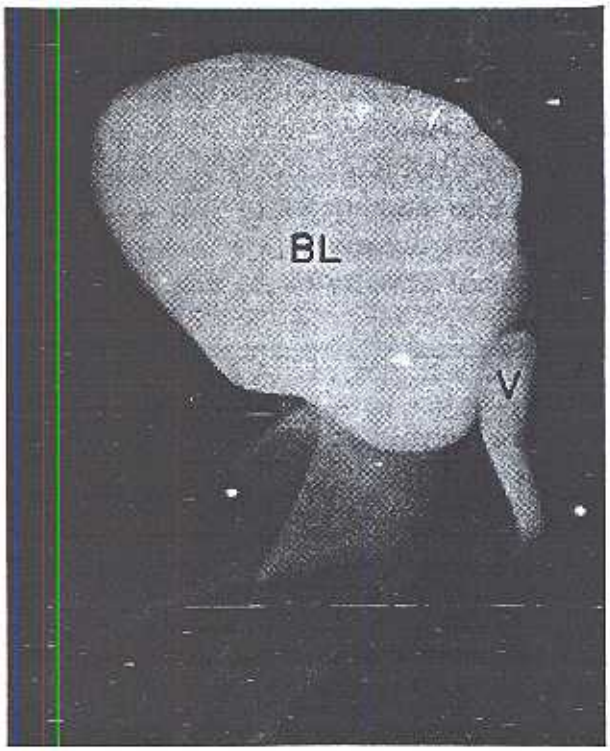

FIG. 2. A genitogram showing contrast material filling the bladder (BL), vagina (V) and uterus (arrow).

males showed false positive results. The results are comparable with those of Bryan *et al.*[5] and Nussbaum *et al.*[11] Our experience confirms that ultrasound is an efficient, rapid and simple procedure. In comparison, genitography revealed a comparable yield of 83% in females but provided more information on the vagina.[4,9,10,12] However, procedural difficulties are sometimes encountered, unlike in sonography. In six (11.3%) of our females in whom the external genitalia were severely virilized, the genitogram failed to show the vagina or uterus: instead, it showed a male-type urethra. Nevertheless, this finding is of considerable clinical importance and reflects the positive correlation with the severity of virilization of the external female genitalia. The combination of ultrasound and genitogram, however, was more sensitive and predicted the presence of a uterus with or without a vagina in 52 (98%) of the females. In one of our patients with 21-hydroxylase deficiency congenital adrenal hyperplasia, who was

initially assigned male sex at his local hospital owing to severe virilization of the external genitalia, ultrasound gave a false negative result and the genitogram showed a male-type urethra. The patient's results are comparable with those of a male pseudohermaphrodite. However, the karyotype revealed 46XX, and Müllerian structures were identified during laparoscopy.

In conclusion, although real time pelvic ultrasonography is less invasive than genitography, its yield in elucidating the genital anatomy is still comparable. Genitography, in addition, reveals further details of the vaginal structures. The combination of both diagnostic modalities is the most informative and gives a better yield.

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