ISSN: 2319-6505

Available Online at http://journalijcar.org

International Journal of Current Advanced Research
Vol 4, Issue 8, pp 261-263, August 2015

International Journal of Current Advanced Research

ISSN: 2319 - 6475

RESEARCH ARTICLE

PELVIC ULTRASONOGRAPHY (US) IN DISORDERS OF SEX DEVELOPMENT (DSD) (AMBIGUOUS GENITALIA)

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A R T I C L E I N F O

Article History:

Received 20th, July, 2015
Received in revised form 31th, July, 2015
Accepted 15th, August, 2015
Published online 28th, August, 2015

Key words:

Ambiguous Genitalia, Disorders Of Sex Development (Dsd), Pelvic, Ultrasonography, Initial, Assessment

A B S T R A C T

Background: Disorders of sex development (DSD), formerly known as ambiguous genitalia, is a birth defect where the outer genitalia do not have the typical appearance of either a boy or a girl.
Aim and objective: To evaluate the role of pelvic ultrasound (US) in assessing the ambiguous genitalia.
Design and setting: A retrospective, hospital based study was conducted at King Khalid University Hospital (KKUH), Riyadh, Saudi Arabia during the period January 1989 and December 2014.
Methods: Medical records of children with the diagnosis of disorders of sex development (DSD) were retrospectively reviewed. Data included age, sex, family history, clinical presentation, and results of relevant laboratory and radiological investigations.
Results: During the period under review (January 1989 and December 2014), a total of 119 patients with ambiguous genitalia were seen. Sixty-three were 46XX DSD and 56 patients were 46XY. Five patients were excluded from the study, due to incomplete procedures, 46XX DSD in three and two with 46XY DSD. The presence of a uterus was predicted by ultrasound in 55 (92%) of 46XX DSD, while no false positive in 46XY DSD.
Conclusion: Pelvic ultrasonography (US) remains the initial modality in assessing ambiguous genitalia, not only because its sensitivity and specificity in identifying the internal organs, but also due to its ability in detecting the enlarged adrenal glands in congenital adrenal hyperplasia which is a common cause.

INTRODUCTION

Disorders of sex development (DSD), formerly termed ambiguous genitalia or intersex conditions, are among the most fascinating conditions encountered by the clinicians. The ability to diagnose these conditions has advanced rapidly in recent years. There are several causes of ambiguous genitalia. In genetic females, congenital adrenal hyperplasia was the commonest. Prenatal exposure to androgens is also a known cause. In genetic males, this includes congenital adrenal hyperplasia and abnormalities in the testes and testosterone metabolism.[1-9]

The pediatric radiologist constitutes an important component of a multi-disciplinary team and plays an important role in defining the genital anatomy. Pelvic ultrasonography can be performed initially. It can be performed at the bedside, and allows visualization of neonate's adrenal glands which may be enlarged in congenital adrenal hyperplasia, however, normal adrenal glands on ultrasonography do not exclude CAH. Ultrasonography is also candemonstrate internal organs. The procedure is a non-invasive, lack of ionizing radiation, no need for sedation, provides high resolution images, most inexpensive and widely available. However, it is important to note that skilled sonographer with the proper probes used in the procedure is quite essential.[8,10-14]

This article aims to evaluate the role of pelvic ultrasound in the assessment of disorders of sex development (DSD) or ambiguous genitalia in a referral hospital at King Khalid University Hospital (KKUH), Riyadh, Saudi Arabia over more than two decades (January 1989 to December 2014).

MATERIALS AND METHODS

The study population consisted of all patients presented or born at King Khalid University Hospital (KKUH), Riyadh, Saudi Arabia over a 25 year period (January 1989 - December 2014). King Khalid University Hospital is the main teaching hospital of King Saud University, and considered as one of the major referral hospitals in the central province of Saudi Arabia. The hospital provides primary, secondary, and tertiary

health care services for the local population and also received patients referred from all over the country. Ambiguous genitalia was considered when there was difficulty in determining the sex of patient on initial examination or when the external genitalia showed significant structural deviation from normal in an apparent male or female.

The karyotyping and appropriate diagnostic, radiological[10] or hormonal,[15-18] evaluation were performed for all patients in the study when indicated.

Medical records of patients were restrospectivelyreviewed and included age at presentation, chromosomal sex, clinical presentation, family history and the appropriate diagnostic (radiological and hormonal) tests.

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 uterus. The ovaries were more difficult to identify with certainty than the uterus, and were therefore not relied on. 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 in their bladder, but the studies were performed after sufficient urine had been allowed to accumulate.[11]

RESULTS

During the period under review, January 1989 and December 2014; a total of 119 patients were seen and evaluated at the endocrine service of King Khalid University Hospital (KKUH), Riyadh Saudi Arabia. This included; chromosomal karyotyping, hormonal investigations and various radiological, (ultrasonography, genitography or magnetic resonance imaging (MRI)) investigations. The aetiological causes were shown in Table 1. Congenital adrenal hyperplasia was the commonest (97%) in 46XX DSD, while a diversity of causes were seen in 46XY. Five patients (three in 46XX DSD and two with 46XY DSD) were excluded from the study, as they had incomplete studies. In female pseudo hermaphroditism (46XX DSD), the presence of a uterus, figure 1 with or without vagina was predicted in 55 (92%) patients by pelvic ultrasonography, however, in male pseudohermaphroditism (46XY DSD) there was no false positive studies by ultrasonography. An enlarged adrenal glands with the cerebriform appearance in abdominal ultrasound was noted in six patients with congenital adrenal hyperplasia, figure 2.

Table1 Aetiology of ambiguous genitalia in 119 patients

Karyotyping	Diagnosis	No. of patient	%
46XX DSD	Congenital adrenal hyperplasia		61 (51.3%)
	21-~-hydroxylase deficiency	51 (48 SW)	-
	11-β-hydroxylase deficiency	9	-
	3-β-hydroxysteroid dehydrogenase deficiency	1 (1 SW)	-
	Isolated clitoromegally		2 (1.7%)
46XY DSD	Androgen insensitivity		
	complete	11	16(13.4%)
	partial	5	
	5-α-reductase deficiency		9 (7.6%)
	Congenital malformation (dysmorphism)		
	local anorectal	4	14 (11.8%)
	generalized	10	
	Extreme prematurity		1 (0.8%)
	Congenital adrenal hyperplasia (due to 3-β-hydroxysteroid dehydrogenase deficiency)		5 (4.2%)
	Hypogonadotrophin deficiency		4 (3.4%)
	Ovotesticular 46,XY, DSD		1 (0.8%)
	Hypospadius		3 (2.5%)
	Persistence of Mulleniar ducts syndrome		1 (1.8%)
	46, XY DSD due to abnormalities of gonadal development	1	2 (1.7%)
	Swyer syndrome	1	
	Denys-Drash syndrome		

SW - salt-wasting
DSD – disorders of sex development

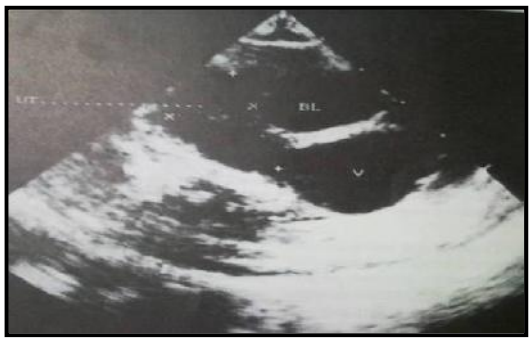

Figure 1 Pelvic ultrasonography of a patient with 46XX DSD, showing a uterus. She was diagnosed with congenital adrenal hyperplasia, 21-α-hydroxylase deficiency

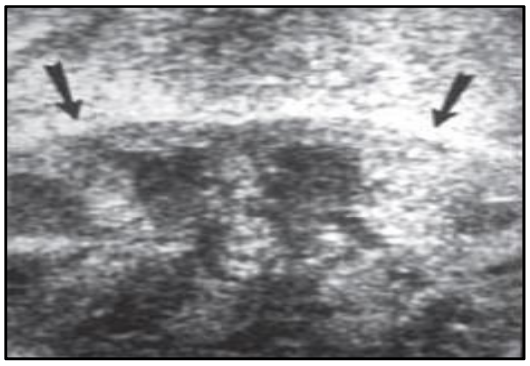

Figure 2 Abdominal ultrasonography, showing enlarged adrenal glands, with the typical cerebriformappearance. She was diagnosed to have congenital adrenal hyperplasia due to 21-α-hydroxylase deficiency

DISCUSSION

Disorders of sex development (DSD), formerly known as ambiguous genitalia, are congenital conditions in which the development of chromosomal, gonadal, or anatomic sex is atypical, and can be classified into four different types; female pseudo hermaphrodites (46XX DSD), male pseudo hermaphrodite (46XY DSD), true hermaphrodites (ovotesticular DSD), and pure gonadal dysgenesis.[1-2] A chromosomal karyotype should be done in all patients. Congenital adrenal hyperplasia is the commonest cause of genital ambiguity in 46XX DSD.[4,5] Imaging plays an important role in depicting the internal organs, therefore, the pediatric radiologist constitutes an important component of a multi-disciplinary team required to work up and manage patients with ambiguous genitalia.[5,6,8] Pelvic ultrasonography is the initial modality for the evaluation of the internal organs.[11,13]

The most common cause of disorders of sex differentiation is CAH,a clue to diagnosis is usually the presence of ambiguous genitalia. In the postnatal period, to assess anatomy of the pelvis, ultrasonography is usually the initial choice.[11]It is easily performed and must include images of the pelvic, scrotum, inguinal, perineal, renal and adrenal areas. The main purpose of the pelvic ultrasonography is to depict accurately the size and morphology of the mullerian structures, the uterus, the vagina and the gonads. In addition to the adrenal glands, the presence of a uterus in a patient with ambiguous genitalia indicates the diagnosis of most likely CAH. The presence of cerebriform pattern is specific for CAH.[14]

Radiological evaluation of internal genital organs constitutes an important component of multi-disciplinary team approach requires in the management of patients presented with ambiguous genitalia. In clinical practice, abdomino-pelvic ultrasonography is the preferred initial imaging modality. Apart from deficiency the uterus, it also may give information on the adrenal glands. However, some cases may necessitate further radiological imaging such as genitography, computerized tomography, or magnetic resonance imaging.

Acknowledgement

The authors would like to thank Ms. Loida M. Sese for typing the manuscript and extend their thanks and appreciation to Mr. Mooath N. Al Jurayyan for his help in preparing the manuscript.

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