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DIAGNOSTIC IMAGING OF DISORDERS OF SEX DEVELOPMENT (DSD) IN INFANTS AND CHILDREN

Radiology

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ABSTRACT

Various disorders of sex development (DSD), result in an abnormal development of genitalia formerly known as ambiguous genitalia or intersex disorders. It Is a complex social and medical emergency. The participation of a multidisciplinary team, including a pediatric radiologist, is needed to address the patient's medical need. The first step in the management of DSD is to assign the sex of rearing, which should be based on multiple factors, such as genotype, the presence, location and appearance of the reproductive organs. various modalities of radiological imaging are used and shown to be of value in diagnosis. Ultrasound (US) is the primary tool for demonstrating internal organs, while genitography is used to assess the uterus, vagina and any fistulas or Complex tracts. Magnetic resonance imaging (MRI) may provide additional and accurate information about the internal gonads and genitalia

KEYWORDS

Ambiguous genitalia, Disorders of sex development, Genitography, Magnetic resonance Imaging (MRI), Radiological Imaging, Ultrasound.

INTRODUCTION

Disorders of sex development (DSD), formerly termed ambiguous genitalia or intersex disorders, refers to a spectrum of congenital conditions in which the development of chromosomal, gonadal or anatomic sex is atypical. It is among the most fascinating conditions encountered by the practicing pediatricians. It constitutes a major complex medical and social emergency requiring a multidisciplinary team approach. Data on incidence and prevalence of DSD is limited. The estimated prevalence is approximately 0.1 % world-wide (ie, 1 in 5000 live birth). This is even higher in a community with an increased rate of consanguineous mating.(1-8)

It is important to decide the appropriate sex of rearing early in life, to avoid the major consequences. The pediatric radiologist plays an important role in evaluating the internal organs and urogenital anatomy in children with DSD. Various modalities of radiological imaging are utilized, including ultrasound, genitography and Magnetic resonance imaging.(1-10)

In this short review we highlight and discuss the different diagnostic radiology imaging being having managed more than 250 patients over three decades at King Khalid university hospital, King Saud University Riyadh, Saudi Arabia.

Diagnostic Radiological (Imaging) Studies

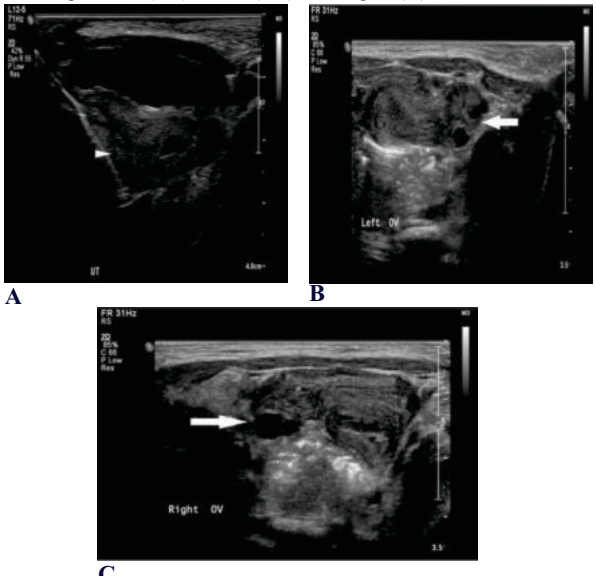
Establishing the genetic sex (karyotype) should be the first step, coupled with determining the anatomical status of the internal organs. Further specific hormonal investigations and therapeutic trials need to be undertaken to specify the cause and, hence, the appropriate therapy. The paediatric radiologist playssignificant and important role elucidating the existing anatomy by utilizing the various radiological tools.

- Ultrasound (US)
- Fluoroscopy-Genitography
- Magnetic Resonance Imaging (MRI)

Ultrasonography

Ultrasound (US) is a non-invasive portable imaging technique that uses sonic energy to produce diagnostic images. Sedation is rarely needed to complete an examination. The images produced provide information concerning the size, shape, and internal structures. Ultrasonolgraphy remains the primary modality for establishing the presence or absence of the uterus. It does not expose the patient to radiation. An ultrasound examination should include the inguinal, perineal, renal, and adrenal. In Ultrasound , identification of the uterus was found to exceed 90% while the ovaries have been seen in 40%(Fig 1-5).

Figure 1 an ultrasound of a newborn baby with ambiguous genitalia caused by congenital adrenal hyperplasia, 21-hydroxylase deficiency, showing bladder (BL), uterus (UT), and vagina (V).



A **B**

C

Figure 2 (A, B & C) Ultrasound pelvis demonstrates presence of the uterus(arrowhead) and both ovaries (arrows).

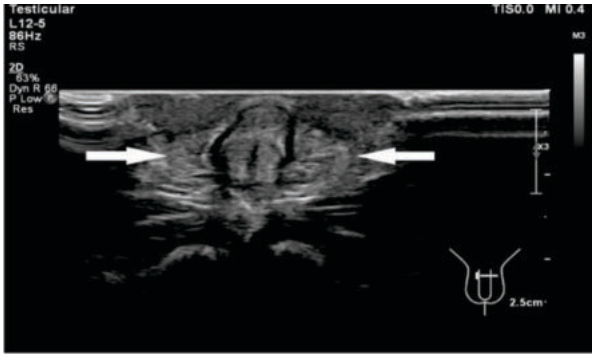


Figure 3 Ultrasound scrotum demonstrats absent testes (arrows).

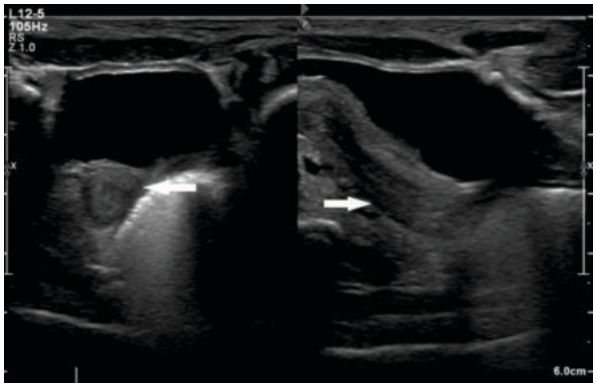
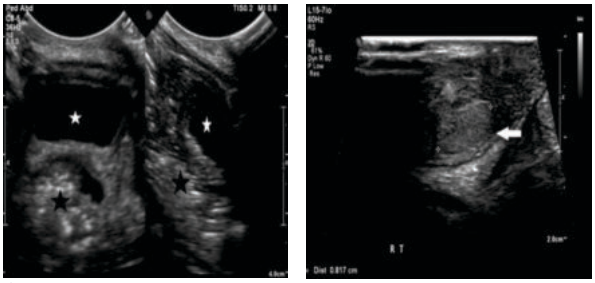


Figure 4 Ultrasound pelvis demonstrates presence of the uterus. No ovaries where visualized.

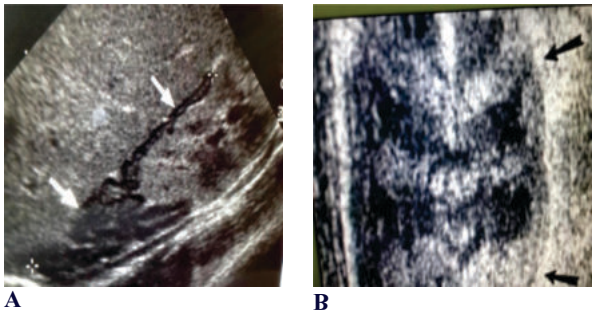


A B

C

Figure 5 Ultrasound pelvis (A) and scrotum (b) and (C) demonstrate absence of uterus and ovaries with presence of both testes in the scrotum (white arrows) with mild hydrocele on left side (white star). The urinary bladder (white star) and rectum (black star).

In some studies, Adrenal glands can be visualized with a limb over 20 mm long and 4 mm wide, with normal cortico-medullary differentiation are suggestive of congenital adrenal hyperplasia (CAH) (Figure 6). However, the presence of normal-sized adrenal glands will not exclude the diagnosis. It is believed that it is not just the size of the gland, but a combination of a limb with greater than 4 mm, a lobulated surface (cerebriform appearance) (Figure 6 A,B), and increased enchogenicity are suggestive the diagnosis of CAH. AL- Alwan associate found a combination of a limb greater than 4 mm, a lobulated surface and stippled echogenicity with a sensitivity of 92% and a speicificity of 100%. (17-19)



A B

Figure 6 Ultrasound images of adrenal glands showing an enlarged, lobulated with maintained normal corticomedullary differentiation (A). It has a "cerebriform" appearance (B) in a newborn with congenital adrenal hyperplasia due to 21 hydroxylase deficiency.

Fluoroscopy-Genitography

This helps determining ductal anatomy. A catheter can be inserted into the distal urogenital sinus. It is more invasive and exposes the patient to much radiation.

Genitography demonstrates a male or female type urethral configuration and any fistulous communication with the vagina or rectum [Figures 7-9]. An adequate genitogram should help identifying the exact location where the urethra and the vagina are joined. These anatomic characteristics are very important for mapping surgical strategy.(20-22)

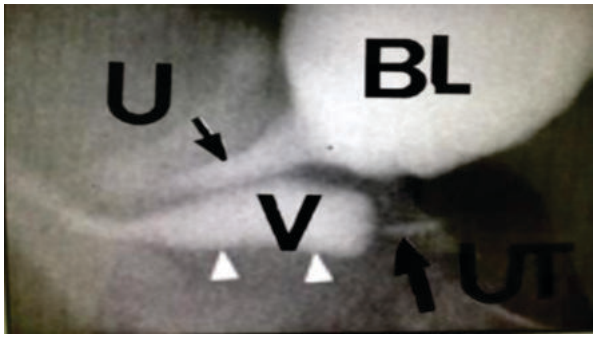


Figure 7 A genitography of a newborn baby with ambiguous genitalia due to 21 hydroxylase deficiency congenital adrenal hyperplasia showing contrast material filling the urethra, bladder (BL), vagina (V) and uterus (U) and cervix.

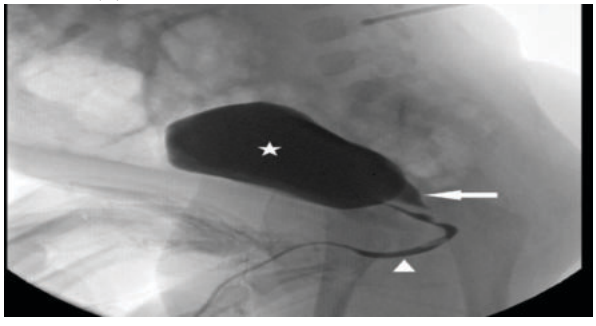


Figure 8 Fluoroscopy genitogram demonstrates the urethra (arrowhead) as a common track filling the vagina (arrow) and the urinary bladder (star).

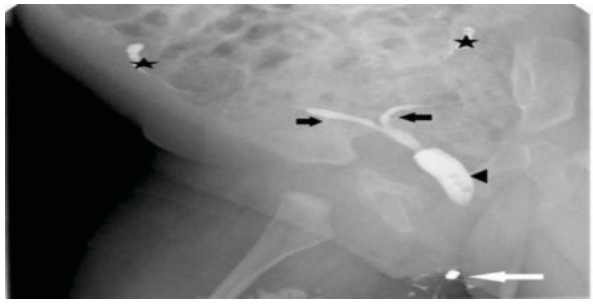


Figure 9 Fluoroscopy genitogram demonstrates contrast filling the

upper vagina (black arrowhead), bicornuate uterus (black arrows) and both fallopian tubes (black stars). The lower vagina was absent with the urinary bladder and urethra were not opacified by contrast despite presence of a common urogenital opening (urogenital sinus) (white arrow).

Magnetic Resonance (MR)

MR has become an accepted modality for diagnostic imaging throughout the body. T1 and T2 weighted MR imaging sequences with their multiplanar capability superior tissue characteristics can provide detailed anatomic information. MR imaging was found useful in evaluating ambiguous genitalia with detecting the uterus in 93% of cases, the vagina in 95%, the penis in 100%, the testis in 88%, and the ovary in 74% (23-25) (Figures 10-13).

MR imaging and US are considered equally sensitive in the evaluation of intrapelvic structures. However, MR imaging is more sensitive than US in detecting the gonads. (18,24,25)

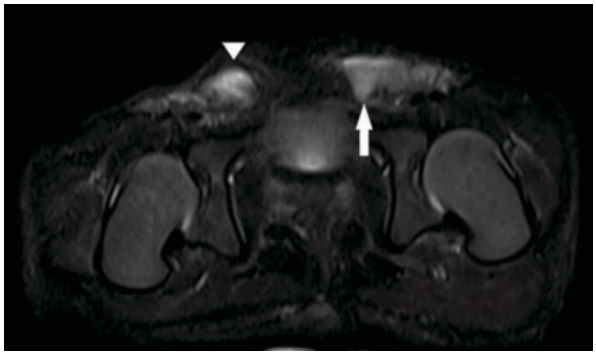


Figure 10 Axial T2 weighted image with fat saturation of the pelvis demonstrates presence of the left testis in left inguinal canal (arrow). The right testis was not visualized with minimal fluid seen in right inguinal canal (Arrowhead).

Figure 11 Sagittal T2 weighted image of the pelvis demonstrates presence of the uterus (star), upper vagina (arrowhead) and clitromegally (arrow). The ovaries and lower vagina were not visualized.

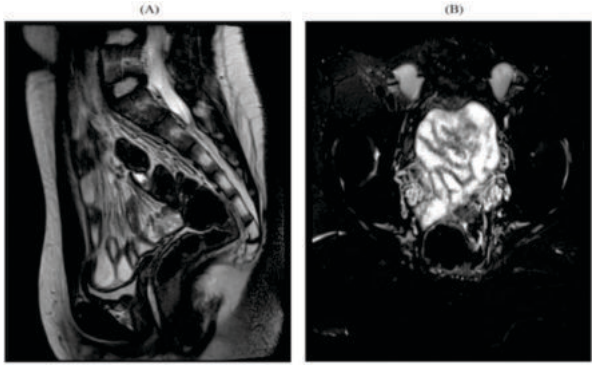
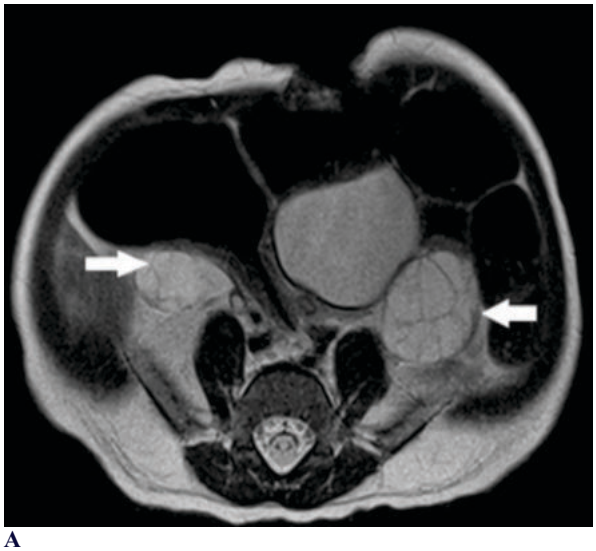
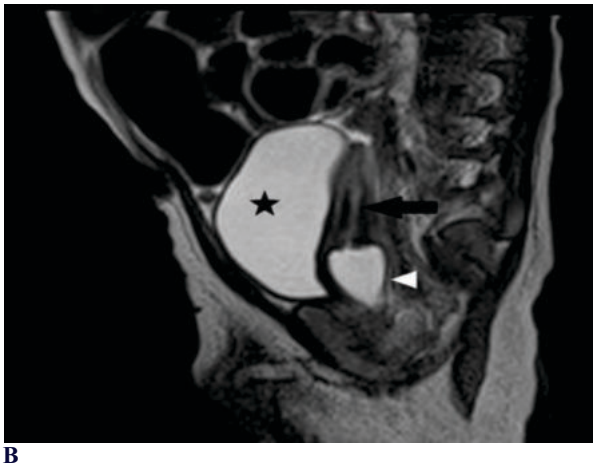


Figure 12 A T2 weighted magnetic resonance imaging (MRI) of the pelvis (a) showing no uterus and (b) showing testicles within the inguinal canals in a patient with complex androgen insensitivity syndrome (CAIS).



A



B

Figure 13 A & B Axial (A) and sagittal (B) MRI pelvis T2 weighted images demonstrates presence of both ovaries with multiple follicles (white arrows), the uterus (black arrow), and fluid filed upper vagina (arrowhead). The lower vagina was not visualized. The urinary bladder (star)Furthermore, those diagnostic tools can even help in diagnosing the primary etiology. (Figures 14-17)

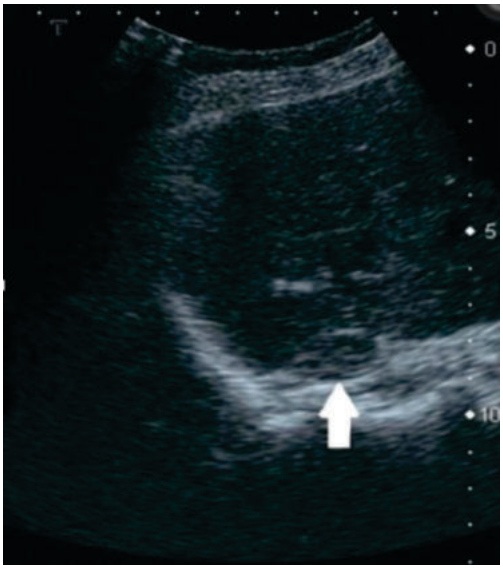


Figure 14 Ultrasound image of the adrenal gland demonstrate adrenal cortex adenoma. (White arrow)

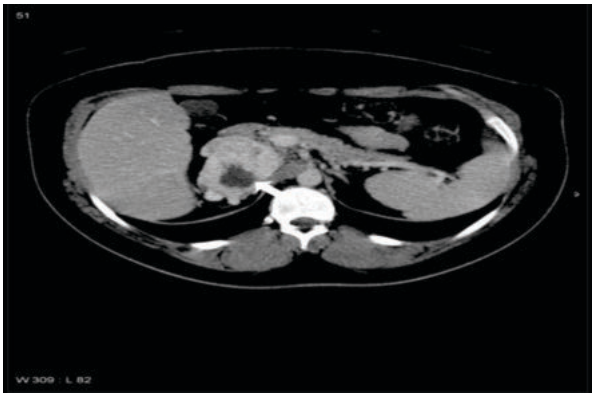


Figure 15 A computed tomography (CT) scan of the adrenal glands showing adrenal carcinoma. (White arrow)

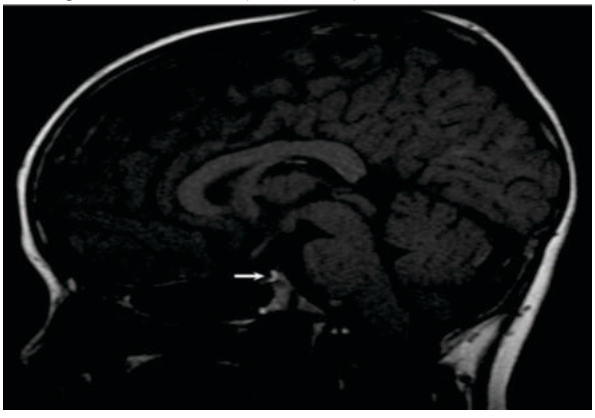


Figure 16 Sagittal T1 weighted Magnetic resonance imaging in a 1-year- old boy with multiple pituitary hormone deficiency (MPHD) showing a small anterior pituitary with bright spot of the posterior pituitary in a normal position

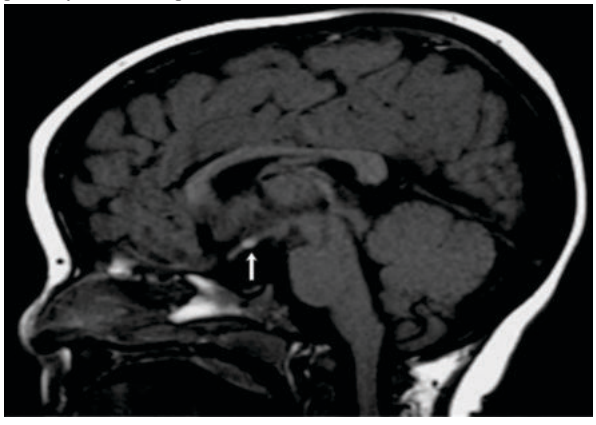


Figure 17 Sagittal T1 weighted Magnetic resonance imaging in a 1.5-year-old girl with multiple pituitary hormone deficiency (MPHD) showing a small anterior pituitary, absent stalk, and ectopic posterior pituitary.

In Conclusion ultrasonography (US), remains the first modality of choice for screening patients with DSD. In addition to elucidating the uterus, it can also give more information on the adrenal. Genitography ,although, less specific and sensitive than ultrasound it provide more information on fistulas and complex tracts within the internal genitalia. MRI is very sensitive and specific in elucidating the internal gonads and genitalia and should be reserved for the complex structures.

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Conflict of interest
The authors have no conflicts of interest to declare.

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