RESEARCH PAPER | Radiology | Volume : 6 | Issue : 3 | March 2016 | ISSN - 2249-555X | IF : 3.919 | IC Value : 74.50

Imaging of A Child with Ambiguous Genitalia

KEYWORDS: ambiguous, hermaphroditism, ultrasound, magnetic resonance

DR. JARVIS PEREIRA
ROOM #110, PRATAPGAD BOYS HOSTEL SMT. KASHIBAI NAVALE MEDICAL COLLEGE AND GENERAL HOSPITAL NARHE PUNE- 411041

DR. SWATI SHAH
FLAT #4, SAYALI APARTMENT LANE #7, PRABHAT ROAD DECCAN PUNE- 411004

DR. SANTOSH KONDE
290/25 PCNTDA, NIGADI, PUNE- 411044

ABSTRACT *Sexual development disorders are a spectrum of congenital conditions characterized by atypical development of sexual organs both anatomically and at the chromosome level. Based on gonadal histologic features, they can be classified into four categories: female pseudohermaphroditism (46XX; 2 ovaries), male pseudohermaphroditism (46XY; 2 testes), true hermaphroditism (both ovaries and testes) and gonadal dysgenesis, either mixed (testes and a streak ovary) or pure (bilateral streak ovaries). The primary modality for demonstrating the internal organs is ultrasonography while magnetic resonance imaging (MRI) is used as an adjunct imaging modality. A team approach involving pediatric endocrinologist, geneticist, urologists and radiologists is required for appropriate gender assignment and allowing timely diagnosis and management. We describe a case study of ambiguous genitalia presented to our department evaluated with multiple imaging modalities.*

CASE REPORT-

A 14 year old girl presented with primary amenorrhea. On clinical examination, both breasts were not developed (Tanner stage II) with masculine built and male pattern voice.

Laboratory examination revealed elevated follicle stimulating hormone (34.44 mIU/ml) and Luneinising hormone (8.28 mIU/ml) and a Karyotype of 46XY (Figures 1 & 2)

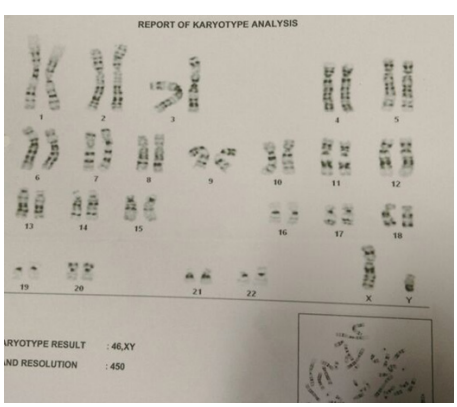


Figure 1- Report of Karyotype analysis revealing result as 46 XY

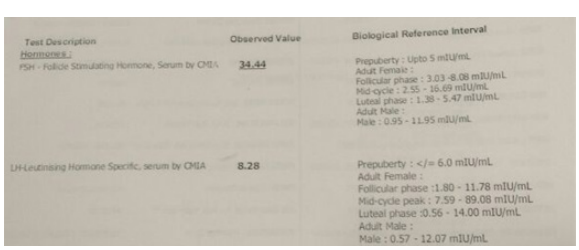


Figure 2- Hormonal analysis revealing elevated FSH and LH levels.

On Ultrasonography, uterus and ovaries were not visualized. Two well defined ovoid structures were visualized bilaterally , adjacent to the labial fold on the left (Figure 3) and in the inguinal region on the right (Figure 4). A prominent phallus (Figure 5) was also noted which on ultrasound showed presence of an artery on color Doppler (Figure 6).

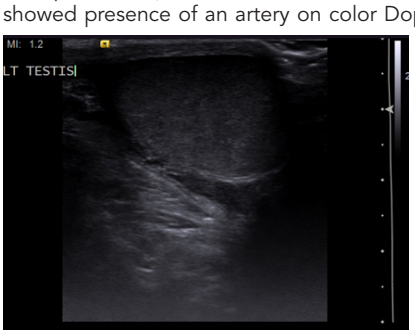
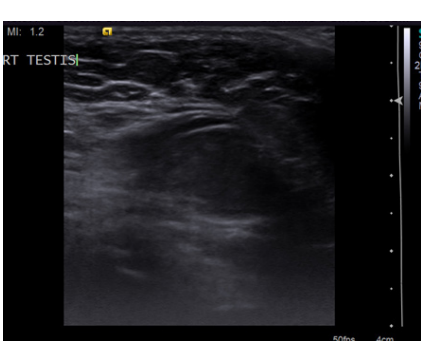


Figure 3- Sonographic image revealing left testis located adjacent to the adjacent labial fold on the left side.

Figure 4- Sonographic image revealing right testis in the inguinal region on the right side.



Volume : 6 | Issue : 3 | March 2016 | ISSN - 2249-555X | IF : 3.919 | IC Value : 74.50

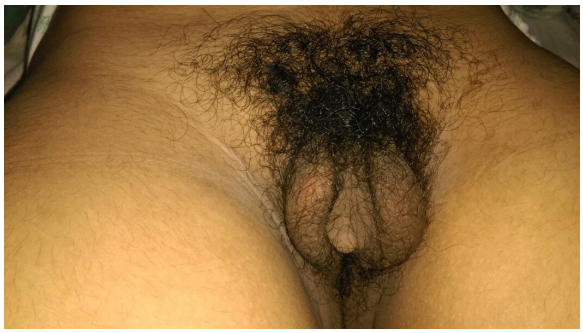


Figure 5- Clinical profile picture showing enlarged phallus.

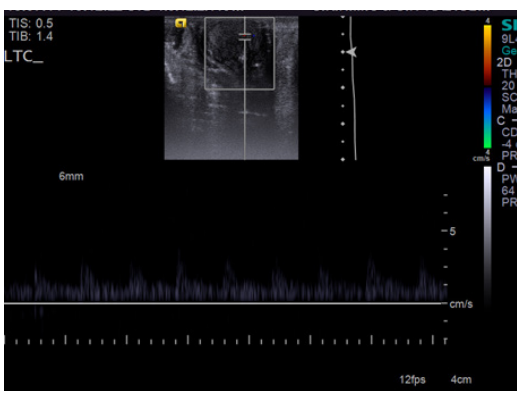


Figure 6- Color Doppler – revealing arterial waveform in a prominent vessel in the phallus.

On MRI, the above findings were confirmed with absence of uterus and ovaries and presence of well defined ovoid structures on either side of midline (Figures 7 and 8).

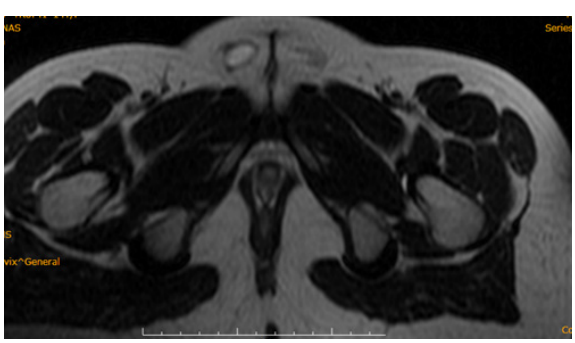


Figure 7- Axial T2WI showing right testis in right inguinal region

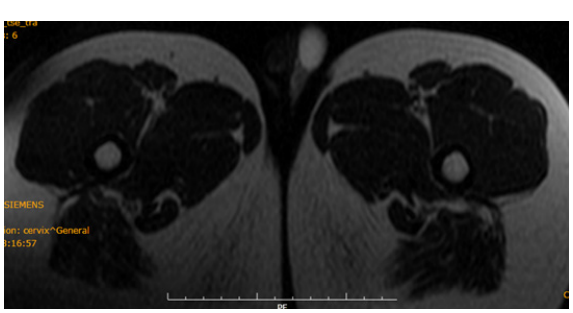


Figure 8- Axial T2WI showing left testis.

Written informed consent in patient's vernacular language was taken.

DISCUSSION-

The chromosomal basis for sex is determined at conception. The important precursors of the genital system are the germ cells, the genital ridge, and the two sets of internal sex ducts, namely, the Mullerian-Paramesonephric ducts and the Wolffian and Mesonephric ducts [1]. The cephalad ends of the paramesonephric ducts open into the peritoneal cavity and form the fallopian tubes while the caudal portion fuses to form the uterus and the cervix [2]. When one mullerian duct fails to develop, a unicornuate uterus results. However, complete failure leads to absence of fallopian tubes, uterus, cervix and part of vagina [3]. Disorders of sexual differentiation (DSD) are classified into 4 different types: Female Pseudohermaphrodites (46, XX DSD), Male Pseudohermaphrodites (46, XY DSD), True hermaphrodites (ovotesticular DSD) and Pure gonadal dysgenesis [4]. Congenital Adrenal hyperplasia is the commonest cause of genital ambiguity in female pseudohermaphrodites [5].

The primary modality for establishing the presence or absence of gonads is US. One ovary has been identified in 40% of patients and in 16% neither ovary has been identified [6].

Though both MR imaging and US are considered equally sensitive, MR imaging is considered more sensitive in detecting gonads [7]. T1- and T2-weighted MR imaging sequences can provide detailed anatomic information.

Ectopic gonads, testes, and non-cystic immature ovaries show intermediate signal intensity on T1-weighted and high signal intensity on T2-weighted MR images with an intermediate-signal-intensity outer rim [8]. Streak gonads are difficult to detect and are seen as stripes of low signal-intensity on T2-weighted images [9]. Streak gonads should be removed as 20-30% children with pure gonadal dysgenesis and 15-20% children with mixed gonadal dysgenesis have a tendency to develop a gonadal neoplasm in either the 1st or the 2nd decade of life [10]. A part of the Y chromosome has been implicated in the development of a malignant neoplasm [11]. High signal-intensity

foci in these gonads could represent neoplastic change [8]. Clitoral hypertrophy in XX DSDs can be differentiated on MR imaging from the penis on the basis of absent or poorly developed supporting penile structures

such as the bulbospongiosus muscle and transverse perinei muscles [12].

Most individuals with 45X/46XY karyotype have a normal male phenotype and escape detection indicating the existence of a bias towards those with clinically evident abnormalities [13,14].

A team comprising of a pediatrician, a pediatric endocrinologist, a urologist, a geneticist, a psychiatrist and a radiologist is needed for the timely evaluation and treatment of children with DSD [15].

CONCLUSION-

Radiological evaluation of the gonads and internal genital organs requires a multidisciplinary team approach in the work-up and treatment of patients with ambiguous genitalia. A multidisciplinary approach must be established for

Volume : 6 | Issue : 3 | March 2016 | ISSN - 2249-555X | IF : 3.919 | IC Value : 74.50

the patient and family considering the psychosocial aspects also.

CONFLICTS OF INTEREST- NONE
FUNDING- NOT REQUIRED

ACKNOWLEDGEMENTS-
I would like to thank **Dr Santosh Konde** for providing me the necessary insight to evaluate the case presented in our institution and encourage me to publish this case. I would also like to thank **Dr Swati Shah** for always being there as a constant support and a gem of a colleague to work with. My gratitude also goes out to my guide **Brig. Dr Hariqbal Singh** for always being a constant source of support for the entire faculty.

REFERENCES-

1. Kucinskas L, Just W. Human Male Sex Determination and Sexual Differentiation: Pathways, Molecular Interaction and Genetic Disorders. Medicina (Kaunas). 2005; 41(8): 633 ñ 640.
2. Moore KL, Persaud TVN. The Urogenital System. In: The developing Human: Clinically Oriented Embryology. 7 th ed. Philadelphia, Pa: WB Saunders; 2003. 287-328
3. Tarek B, Richard SL. Fallopian Tube disorders. Medscape ñ Web M.D. Professional Update: March 5, 2012. Accessed: March 18, 2012.
4. Wright NB, Smith C, Rickwood AM, Carty HM. Imaging children with ambiguous genitalia and intersex states. Clin Radiol 1995;50:823–829.
5. Lee PA, Houk CP, Ahmed F, Hughes IA. Consensus Statement on Management of Intersex Disorders. Paediatrics. 2006; 118(2): 488 ñ 500.
6. Cohen HL, Shapiro MA, Mandel FS, Shapiro ML. Normal ovaries in neonates and infants: a sonographic study of 77 patients 1 day to 24 months old. AJR Am J Roentgenol 1993;160:583– 586.
7. Mansour SM, Hamed ST, Adel L, Kamal RM, Ahmed DM. Does MRI add to ultrasound in the assessment of Disorders of sex development? Eur J Radiol 2012; 81 (9); 2403-10.
8. GambinoJ, Caldwell B, Dietrich R, Walot I, Kangarloo H. Congenital disorders of sexual differentiation: MR findings. AJR Am J Roentgenol1992;158:363–367
9. HricakH, Chang YC, Thurnher S. Vagina: evaluation with MR imaging. I. Normal anatomy and congenital anomalies. Radiology1988;169:169–174.
10. Coran AG, Polley TZ Jr. Surgical management of ambiguous genitalia in the infant and child. J Pediatr Surg 1991;26:812–820
11. Looijenga LH, Hersmus R, Oosterhuis JW, Cools M, Drop SL, Wolffenbuttel KP. Tumor risk in disorders of sex development (DSD). Best Pract Res Clin Endocrinol Metab 2007;21:480–495
12. HricakH, Marotti M, Gilbert TJ, et al. Normal penile anatomy and abnormal penile conditions: evaluation with MR imaging. Radiology1988;169:683–690.
13. Chang HJ, Clark RD, Bachman H. Chromosome mosaicism in 6,000 amniocenteses. Am J Med Genet 1989;32:506–13
14. Wilson MG, Lin MS, Fujimoto A, et al. The phenotype of 45,X/46,XY mosaicism: an analysis of 92 prenatally diagnosed cases. Am J Hum Genet 1990;46:156–67.
15. Moshiri M, Chapman T, Fechner PY, Dubinsky TJ, Shnorhavorian M, Osman S, et.al. Evaluation and Management of Disorders of Sex Development: Multidisciplinary Approach to a Complex Diagnosis. Radiographics 2012; 32:1509-1018.