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IMAGING OF AMBIGUOUS GENITALIA

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ABSTRACT... Objective: To establish the role of imaging in Ambiguous Genitalia. **Period:** June 2006 to June 2008. **Place:** Radiology Department, Mayo Hospital, Lahore and Allied Hospital Faisalabad. **Patients and Methods:**12 cases referred to us with Ambiguous Genitalia were taken and evaluated with Ultrasound and Magnetic Resonance Imaging. **Results:** Following findings were observed, incomplete scrotal fusion with Ambiguous genitalia was observed in 41.7% infants. Whereas testis were localized in 33.3% by ultrasound & remaining by MRI. Imaging diagnosed that 66.7% were male & remaining female. Lab tests and surgical evidence prove that imaging results were in agreement. **Conclusion:** Imaging has a conclusive role in Ambiguous Genitalia and Ultrasound is the first modality to look for Internal Genitalia.

Key words: Ambiguous Genitalia Ultrasound, MRI, Testis, Uterus.

INTRODUCTION

Newborns with Ambiguous genitalia, is the medical term for a rare conduction in which a newborn's external genitalia don't appear to be clearly either male or female. In Ambiguous genitalia, the baby's genitalia may not be well-formed, or the baby may have general characteristics of both sexes[1]. Ambiguous genitalia can be very upsetting to parents and other family members, both because of the uncertainly involved and because of the social stigma attached to not knowing a child's sex right away. While Ambiguous genitalia can present a difficult and complicated situation, medical advances can take much of the guesswork out of the process of assigning a sex to your child, and corrective surgery can help.

The first indication of Ambiguous genitalia is through physical examination. However, investigation work up that is generally recommended in Ambiguous genitalia includes blood and urine tests to measure hormone levels, chromosome analysis in order to determine the genetic sex (XX or XY), ultrasound to check for the presence of internal sex organs, a biopsy of newborn's reproductive organs to determine if the organs will produce appropriate sex hormones for the sex assigned to the child and X-ray to see presence and size of a

vagina with a genitogram[2].

Typically, Ambiguous genitalia in genetic females over masculized (babies with XX chromosomes) may have features like an enlarge clitoris that has the appearance of a small sized penis, the urethral opening can be anywhere along, above, or below the surface of the clitoris, the labia may be fused, resembling a scrotum, the infant may be thought to be a male with undescended testes, sometimes a lump of tissue is felt within the fused labia, further making it look like a scrotum with testicles[3].

Whereas, in a genetic male under masculized (XY chromosome), Ambiguous genitalia typically may include the features like a small penis (less than 2-3 centimeters) that resemble an enlarge clitoris, the urethral opening may be anywhere alone, above, or below the penis; it can be placed as low as on the perineum, further making the infant appear to be female, there may be a small scrotum with any degree of separation, resembling labia, and undescended testicles commonly accompany Ambiguous genitalia[4,5].

Imaging has a defined role in Ambiguous genitalia both during intrauterine life and after birth. Ultrasound is the most commonly used imaging investigation for the work up of Ambiguous genitalia and to look for internal reproductive organs. Magnetic Resonance Imaging (MRI) has come as advancement and it has now become very easy to define the internal organs with the help of this tool. Contrast imaging, such as a retrograde genitogram or a voiding cystourethrogram, may demonstrate the anatomy of the urogenital system, the urethra and the vagina, if present. Keeping above facts in view, the present study only included use of Imaging for diagnosis of Ambiguous genitalia in infants[4,6,7].

PATIENTS AND STUDY

OBJECTIVE

To establish the role if imaging in Ambiguous genitalia.

Patients and Methods

Twelve cases referred to us with Ambiguous genitalia were included in this study and their sex was determined with Ultrasound and MRI (Ultrasound machine used was Logic 5 GE with convex 3.5-5 MHz and high frequency Linear probe of 7-10 MHz frequency. MRI was performed on 1.5 Tesla machines.

RESULTS

Incomplete Scrotal fusion with Ambiguous genitalia (Table I) was observed in five out of 12 patients (41.7%) where in tests were localized in four patients by ultrasound and one patient by MRI. Fused Labia and enlarge Clitoris were observed in four out of 12 patients (33.3%). However, their Uterus was identified by Ultrasound. Micro penis with non-palpable testes were seen in three out of 12 patients (25%) and their testes were localized by ultrasound.

Imaging diagnosed that out of 12 patients 8 were male and remaining (four) as females. Furthermore, lab test and surgical evidences proved that imaging results were in agreement.

Table-I. Imaging findings in patients with Ambiguous genitalia.

Clinical presentation of the patients	No. of Pts	%age
Incomplete Scrotal fusion	5	41.7
Fused Labia and enlarged Clitoris	4	33.3
Micro penis with non-palpable testes	3	25.0

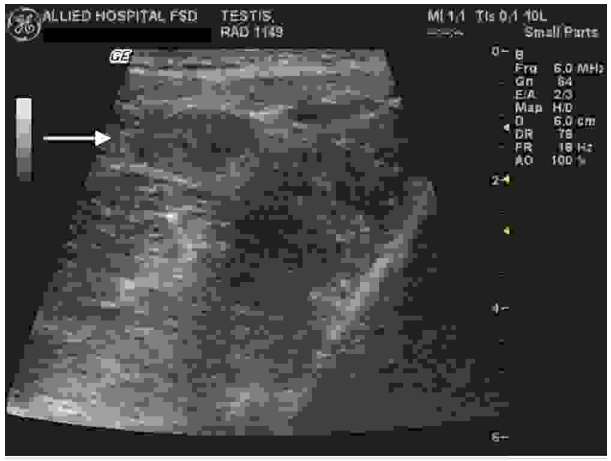

Fig-1. Left undescended testis at level of deep inguinal ring

DISCUSSION

After initial diagnosis of Ambiguous genitalia through physical examination it requires a series of investigations to assign the sex to the newborn. The blood and urine tests to access the hormonal levels require sophisticated laboratories which are not available in towns and cities and they are costly. Chromosomal analysis and biopsy of newborn's reproductive organs are invasive as well as expensive. The investigation like genitogram are reserved for complicated cases. However, ultrasonography to check for the presence of internal sex organs is non-invasive, easily available and first hand to assign the sex of a newborn. Whereas MRI is reserved for difficult cases, expensive, not easily available. In our study we diagnosed 11 out of 12 patients by ultrasonography and 1(one) out of 12 by MRI. The clinical presentation of male patients was incomplete scrotal fusion and micro penis with non-palpable testes. These are present in 8 infants out of 12 with Ambiguous, genitalia, whereas 4 out of 12 were diagnosed as Females that presented with fused Labia and enlarged clitoris. The follow-up of these cases with Lab tests and surgical evidences proved that the Imaging results were in agreement[8,9].

Once investigation for Ambiguous genitalia have been completed and a diagnosis has been made, the sex of the infant may still be questionable. Criteria that must be considered include not only the genetic karyotype, but also the potential to have an unambiguous appearance, the potential to have normal sexual functioning and fertility. Consequently, the sexual changes that will occur at puberty as well as the capabilities of reconstructive surgery and hormonal therapy all play a role in the decision. The child should be reared on a sex where it can lead a full life remembering that the vagina can be created surgically but male organs can not be made surgically.

CONCLUSION

Imaging has a conclusive role because it is cheaper, time saving, easily available and non-invasive techniques (ultrasound and MRI) to diagnose the Ambiguous genitalia. Such techniques should be performed at the initial stage rather than implementing expensive and invasive tests. Ultrasound is being the first modality to look for Internal genitalia followed by MRI.

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