RADIOLOGICAL EVALUATION OF CHILDREN WITH ATYPICAL GENITALIA DUE TO DISORDER OF SEX DEVELOPMENT (46, XY DSD)

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INTRODUCTION

Disorders of sex development (DSD) comprises a heterogeneous group of conditions involving interference with normal sex differentiation and hormonal production in the embryo, Imaging has a very useful role in assessing the patient's phenotypic sex. The radiologist can assist the DSD-management team in identifying the internal genital anatomy and adrenal glands.

AIM

The current study aimed at radiological evaluation of 46 XY DSD cases .

METHOD

All cases of 46, XY DSD who were referred to Endocrinology Clinic in Alexandria University Children's Hospital for evaluation because of atypical genitalia over one year (2019) were included and subjected to clinical assessment including external genitalia using external musculinization score (EMS). All cases had Ultrasound pelvis and inguino-scrotal region to assess internal genital organs, localize the gonads and measure the testicular volume. MRI abdomen and pelvis was done when ultrasound on abdominal and pelvic regions was non-conclusive. Laparoscopy and testicular biopsy for localization of the gonads when they are not found.

RESULTS

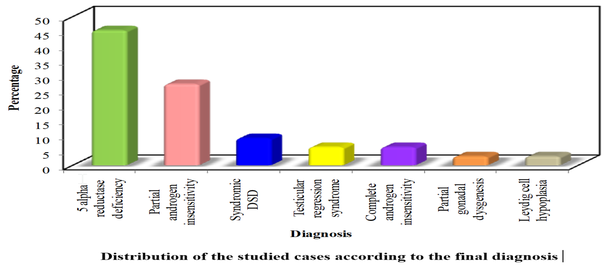
The age of presentation ranged between 0.6– 14 years with a mean of 3.34 ± 1.85 years.

Most of the cases were initially assigned as males (18 cases, 75%).

Five alpha reductase deficiency was the most common diagnosis (12 cases, 50%). While leydig cell hypoplasia were the least common(1 case, 4.1%)

Using ultrasonography, 20 cases had normal testicular volume for age (83.3%). Two cases had bilateral non-detectable gonads either clinically or by ultrasonography, so they had abdominal and pelvic MRI. One of them had male phenotype with bilateral cryptorchidism,and MRI showed one gonad was atrophied and the other was partially atrophied in the inguinal canal. The other case had female phenotype with bilateral cryptorchidism and MRI showed bilateral abdominal testes.

Laparoscopy was done four patients in our study. 2 cases showed bilateral vanishing testes. Third case showed Mullerian duct remnants and bilateral gonads that showed premature testicular tissue by pathological examination. A therapeutic laparoscopy was done for the fourth case that had female phenotype and abdominal testes for gonadal excision at puberty.



Distribution of the studied cases according to the final diagnosis |

: **Distribution of the studied cases according to ultrasonographic testicular volume according to age (n = 23)#**

Testicular Volume	No.	%
Small	4	8
Normal	20	87
Large	1	5

Cases with bilateral undetected gonads were excluded

CONCLUSIONS

The diagnostic management of XY DSD cases remains the greatest challenge.

MRI and laparoscopy are not more sensitive than ultrasonography in the evaluation of gonads. Thus, they can be reserved when ultrasonography is non-conclusive.

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