AICOG 2025
Mumbai
67th All India Congress of Obstetrics & Gynaecology
FOGSI celebrates 75 years - Diamond Jubliee
8-12 January, 2025 | JIO World Convention Centre

Poster Number: EP120 Name: DR PRATHA APRAMITA (PG, O&G)

GUIDE- DR VANDANA MOHAPATRA (ASST PROFESSOR, O&G)

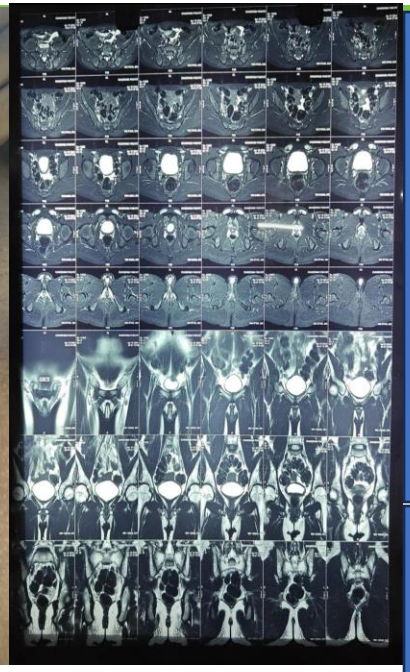
Title: A RARE CASE OF 46XY DSD WITH 17 BETA HYDROXYSTEROID DEHYDROGENASE 3 DEFICIENCY

INTRODUCTION

DSD is an umbrella term referring to a collection of conditions in which chromosomal, gonadal, or anatomical sex development is affected. Its prevalence widely varies from 1 in 5000 to 6000 in general population. They mainly present with ambiguous genitalia, amenorrhoea or infertility in later stage of life.

CASE REPORT

A 18yr individual presented to GOPD with chief complaint of not attaining menarche. On examination- breast- tanner stage 2, Axillary and pubic hair- tanner stage 4. On examination- B/L Firm, smooth and mobile masses of size 3×2cm in inguinal region. On IOV- A Phallus of length 2.5cm seen with EUM at the base of the phallus, A blind vaginal opening of 2cm present. Karyotype was 46XY. Sr LH- 19MIU/ml, FSH- 12.07MIU/ml, testosterone-363ng/dl, T/DHT- 4.65, T/A- 0.36. Her MRI showed B/L undescended testis with no evidence of uterus, ovaries or prostate like structure. She underwent B/L orchidectomy with McIndoe vaginoplasty and clitoroplasty. Estradiol valerate 1mg and spironolactone was advised.



DISCUSSION

17betaHSD3 deficiency is a rare cause of XY undervirilization (1 in 147,000). It should be suspected in females with inguinal hernia or clitoromegaly in infancy or early childhood or virilization at puberty associated with low T/A ratio. Molecular genetic testing helps in confirming diagnosis. Almost half of the patients change from female to male sex but in our case she retained female sex after proper counseling and appropriate surgery.

CONCLUSION

A patient with DSD requires a multidisciplinary approach, inclusion of parents and most importantly assuring the psychological well being of the patient. Urgent genetic testing is advocated in rare cases of DSD as indeterminate hormone results can delay diagnosis and prolong intervention.

REFERENCES

1. Berek JS (eds). Berek and Novak's Gynaecology, 16th edn., Wolters Kluwer, Philadelphia, USA, 2019, p. 270
2. Mendonca BB, Gomes NL, Costa EM, Inacio M, Martin RM, et al. 46,XY disorder of sex development (DSD) due to 17β-hydroxysteroid dehydrogenase type 3 deficiency. J Steroid Biochem Mol Biol. 2017