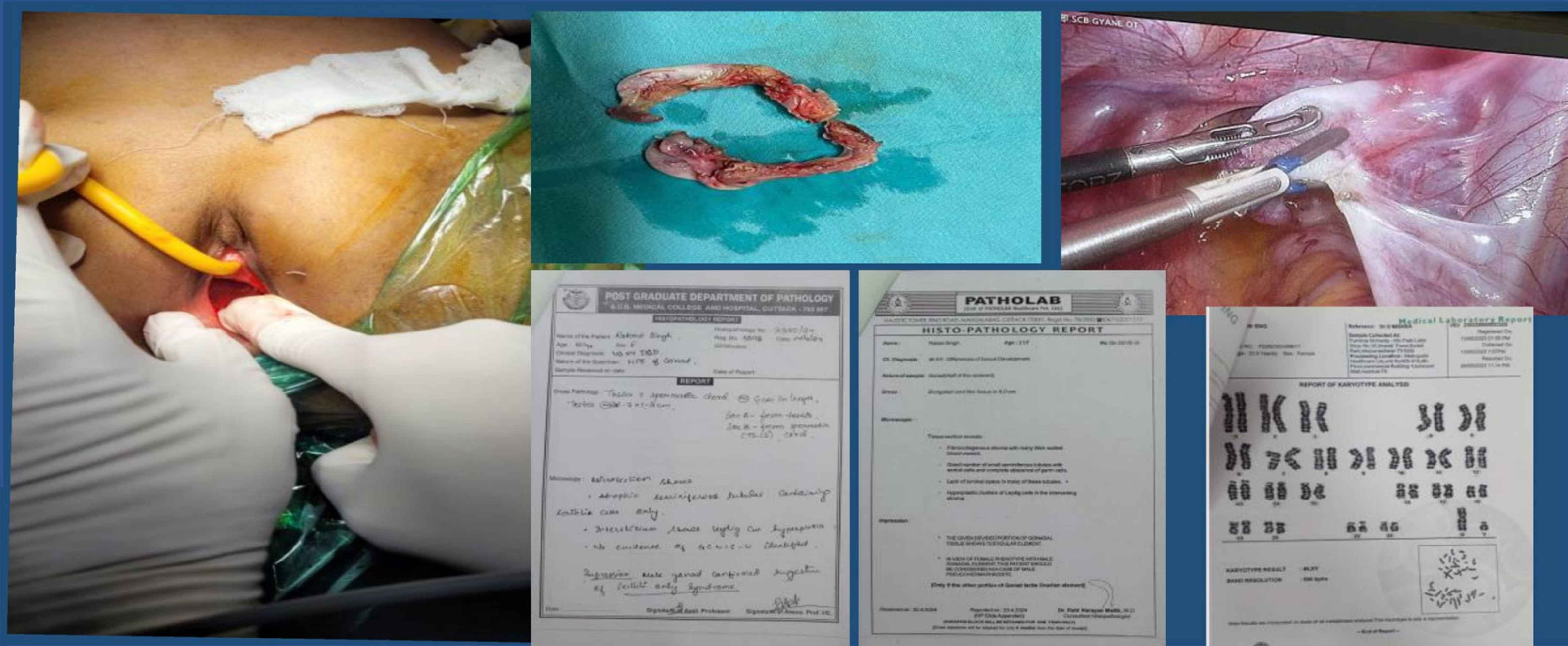


A RARE CASE OF PRIMARY AMENORRHEA,FEMALE WITH 46XY DSD
GUIDE-PROF.DR. K. R. MOHAPATRA[EX-HOD],DR DEEPA SETHI [ASST. PROF.]
DR MEENA TANDI[2ND YEAR PG RESIDENT]
DEPT. OF OBSTETRICS & GYNECOLOGY,SCB MCH,CUTTACK

INTRODUCTION
Disorders of sexual development(DSDs) refers to a group of conditions characterized by atypical development of chromosomal,gonadal, or anatomical sex.These disorder can result in variation in sexual differentiation, leading to ambiguous genitalia or discrepancies between an individual’s genetic sex and their anatomical characteristicsl.

CASE REPORT
A 21 years female admitted to our facility with chief complaints of not attending menarche even after development of thelarche at 14 years and puberche at 19 years. There was No h/o cyclical abdominal pain.

EXAMINATION
Patient vitals stable,Height-144cm,Weight-30kg, BMI-14.5. On local examination;
Breast-Tanner stage 2
Axillary & Pubic hair-Tanner stage 2
Inguinal area-no mass palpable
Inspection of vulva-Vaginal opening present
Per Rectal examination-No uterus No cervix felt



INVESTIGATIONS-
USG-Hypoechoic lesion noted inferior to bladder with 2 cystic elongated structure noted adjacent to it.Uterus & Ovary not visualised.MRI-Rudimentary gonadal tissue S/O of gonadal agenesis.KARYOTYPE-46XY
CBC,ALL Serum-WNL
Sr TSH-1.2,FT3-2.4,FT4-12.4
LH-29.6,FSH-57.8
Sr Prolactin-7.52
Total Testosterone-83.6
Estradiol-<5pg/ml, Vit.D-14.10
2D ECHO-Normal LV Function

TREATMENT
BILATERAL LAPAROSCOPIC GONADECTOMY
Histo-Pathology Shows-SERTOLI ONLY SYNDROME
PLAN-HORMONAL REPLACEMENT THERAPY
(ESTRADIOL VALERATE 2mg)

DISCUSSION-
DSDs can arise from genetic mutations,hormonal imbalance,or environmental factors, and they encompass a range of conditions, including congenital adrenal hyperplasia,androgen insensitivity syndrome,and Turner syndrome.Diagnosis often involves genetic testing,hormone assessments,and imaging studies.

CONCLUSION-
Management of DSDs may include hormone therapy,surgery and psychological support,tailored to individual’s needs &circumstances.Understanding DSDs is crucial for promoting awareness,reducing stigma.

REFERENCE
Speroff, L., Glass, R. H., & Kase, N. G. (2011). Clinical Gynecologic Endocrinology and Infertility (9th ed.). Lippincott Williams & Wilkins.
2018 ESPED (European Society for Paediatric Endocrinology) Guidelines