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Poster Number: EP 104 Name: Dr HARISH MAHARANA

PG Resident, MKCG MCH,Berhampur,ODISHA

Title: DISORDERS OF SEXUAL DIFFERENTIATION - CONGENITAL ADRENAL HYPERPLASIA

INTRODUCTION

•Disorders of sexual development (Dsd) or differentiation is a congenital condition in which development of chromosomal, gonadal and anatomical sex is different.
•Most common cause of 46XX Dsd is virilizing forms of CAH. Most common enzyme deficiency is 21-hydroxylase.
•Incidence at birth -1.2000 –1.4500.
•Challenges of ambiguous genitalia at birth -sex of rearing, parent and patient education and medical management.

CASEREPORT:

•A 28-yr old unmarried adult female patient presented to OPD with abnormal genitalia and excessive facial hair with H/O delayed menarche
•She is a 3rd order child who delivered vaginally out of 3rd degree consanguineous marriage. She was designated as female sex at birth.
•Menstrual history-Menarche at 19 yrs (delayed) PMC-3-4 days /45-50 days ,
•LMP-12 days back

Clitoromegaly with CI-225mm²

INVESTIGATION:

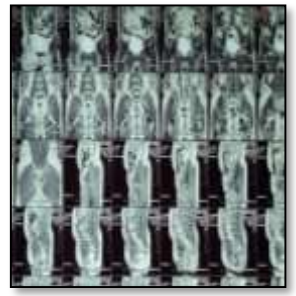
CBC,LFT,RFT and TFT were normal.Karyotype-46XX

S. Na^+ -140 mEq/L, S. K^+ - 4.4 mEq/L
S. Estradiol-112.7 pg/ml, S. Testosterone –186.3 ng/dl
S.LH- 0.6 mIU /ml, FSH- 3.49 mIU /ml, S .cortisol- 10. 6 ug/dl
17OH Progestrone - 231 ng / ml

USG- Right and left ovaries were normal,
Hypoplastic uterus–**6.4x2.2x3.1cm,**ET-**6mm**
On CE-MRI –Diffuse enlargement of B/L adrenal glands (thickness -> **10 mm** , length > **5 cm**), maintained their shape.

Coarse dark pigmented hairs over chin upper lip, upper abdomen and thigh with Breast tanner stage - 3

Post op image following clitoroplasty



CE – MRI showing enlarged adrenal glands

FINAL DIAGNOSIS

Congenital Adrenal hyperplasia (simple virilizing type)

MANAGEMENT

Tab prednisolone 10mg at Morning and 5mg at Night for 7days .Then Tab Prednisolone 5mg at Morning and 2.5mg at Night life long. Patient underwent clitoroplasty and vaginoplasty for enlarged phallus in plastic surgery department, and facial hair reduction with diode laser therapy.

DISCUSSION

- CAH is an autosomal recessive disorder caused by defects in steroidogenic enzymes involved in cortisol and aldosterone synthesis in the adrenal glands. Mutations in CYP21A2 lead to a deficiency of 21-hydroxylase resulting in reduced cortisol, overproduction of pituitary corticotrophin, adrenal hyperplasia and increased adrenal androgens and 17OH.
- The classic form includes two phenotypes: simple virilizing and salt-wasting. All patients with classic 21-OHD exhibit ambiguous genitalia, typically diagnosed at birth but rare cases may present later. Diagnosis is confirmed by high serum 17OH-progesterone levels.
- Management in adults focuses on blocking hyperandrogenism and preventing complications. Hydrocortisone is the preferred glucocorticoid while prednisolone provides stable replacement effects. Spironolactone, an aldosterone antagonist, reduces hirsutism, clitoral size, and supports breast development. For severe virilization, surgical options like vaginoplasty or clitoroplasty are considered.