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RESEARCH ARTICLE

COMPLETE ANDROGEN INSENSITIVITY SYNDROME

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Key words:-

Abstract

1 year old Baby born on 30.05.2012, got admitted in Paediatric surgery , Government Rajaji Hospital attached to Madurai Medical College, Madurai with the history of band on genitalia & bilateral inguinal swelling since birth reared and named as female.

On **examination** Testis like structure noted in both inguinal region.
Urethral and vaginal orifice present.

Karyotyping done showed 46 XY.
The following investigations done.
Serum 17α OH Progesterone 0.70 ng/ ml.
Serum Total Testosterone 144.89 ng/dl.
Serum Electrolytes; Na+ 135 m mol/lit.
K+4.40 m mol/lit.
Cl- 101m mol/lit.

Complete Haemogramdone . Blood sugar 100mg/dl.
Blood urea-39 mg/dl.Creatinine 0.6 mg /ml. Blood group A Rh Positive.TSH level is 1.10 Miu/ml.FSH 4.64, LH level 0.45,

Buccal mucosal smear revealed absence of Barr body.

A provisional diagnosis of **Complete Testicular Feminising Syndrome** made with the **female phenotype and XY Karyotype.**

Bilateral groin exploration with Tunicotomy proceeded. Both Testis visualised in supra marginal region.1 cm x 1cm swelling reducible on palpation. Uterus not seen.

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Bilateral Herniotomy with Testicular Biopsy done. Right inguinal skin crease incision made. Hernial sac identified and separated from cord structures.Testis 1.5 x 1.5 cm present. Longitudinal Testicular biopsy taken and closed with 4-0 vicryl. Testis placed in supra inguinal pouch and closed in layers. Same procedure done in opposite side with same findings.

Biopsy showed testicular parenchyma with immature seminiferous tubules.

MRI showed a small fibrous structure at uterine region (1x 0.34 cm). Testis like structure 1.95 x 1,12 on left side and 1.75 x1.6 on right side.

Discussion:-

Human genome contains about 3billion nucleotide pairs and in diploid cells, they are organised into 23 pairs of chromosomes. X and Y are concerned with determination of sex & are called as sex chromosomes. As stated in **Lyon Hypothesis**, in females, one of the 2 X chromosomes paternal or maternal derived is inactivated during embryogenesis. This inactivation is passed to all the somatic cells, while the germ cell in female remain unaffected. That means ovary will have always active X chromosome. The inactive X chromosome in the somatic cells in females lies condensed in the nucleus as sex chromatin. **Nuclear sexing** is done for genetic females by preparing and staining the smears of squamous cells scrapped from oral cavity or by identifying Barr body attached to the nuclear lobes in the circulating neutrophil in females. A minimum of 30% cells positive for sex chromatin indicates the person as female genetically.

Testicular Feminisation coined by **Morris et al** was applied to a highly distinctive X linked disorder **Grumbach MM** in which affected males are phenotypic females and develop female secondary sexual characters at puberty but fail to menstruate. Affected individuals are genetic males with 46 XY karyotype and have testes. It is a X linked recessive disorder with mutation in AR gene.

Prevalence is between 1 in 20,000 to 1 in 60,000 live male births**Jagiello**. 1% to 2 % of phenotypic females with inguinal hernia have this syndrome.

Androgen insensitivity **Quigley et al** represents the most common cause of male pseudohermophroditism **Bale et al.** Phenotypically they have unambiguous female genitalia, hypoplastic labia majora, a blind vaginal pouch, absent or vestigial mullerian structures, testis located in the labia, inguinal canal or in abdomen.

Hereditary defects in the X linked gene that encodes the androgen receptor cause a spectrum of syndromes of incomplete virilisation in 46 XY men who have testes and male testosterone levels but who are resistant to their own & exogenous androgens. Affected individuals are phenotypic women with Testicular Feminising Syndrome.

The sex of rearing is female in the complete form of androgen insensitivity and the gonads need to be removed owing to a risk of malignancy in 10% -30 % cases.

Conclusion:-

Testicular Feminizing Syndrome described by **Norris**in 1953**VG Padubidr**i is now designated as **Complete or Partial Androgen Insensitivity Syndrome.**The peripheral receptors for testosterone are absent, scanty or fail to respond totestosterone.**MRI** showed a small fibrous structure at uterine region (1cm x 0.34 cm). Testis like structure 1.95 x 1,12 on left side and 1.75 x1.6 on right side with ultra sound revealing absence of uterus and ovaries.Supplementing Oestrogen is required after gonadectomy for the development of the breasts as well as to prevent osteoporosis.

With the consent of parents, an appropriate psycho social support system,counselling done to the child. Parents were informed to rear her as a girl child and even get her married at a later date after a vaginoplasty and may adopt a child as she has lost her fertility.

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