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TURNER SYNDROME –A CLASSICAL CASE

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ABSTRACT

Turner syndrome is the commonest (X linked) Sex Chromosomal Inherited disorder resulting in dwarfism and amenorrhea in female sex .Association of ovarian dysgenesis ,congenital hypergonadotrophic hypogonadism,cardiac anamolies ,endocrine abnormalities with turner syndrome are often seen. The morbidity and mortality rates are high in females with Turner syndrome.

A female adolescent aged 14 years presented with features of growth retardation had the clinical signs of Turner syndrome.Laboratory investigation and Karyotyping analysis confirmed the diagnosis. [1]

Keywords: Amenorrhea , cardiac anomalies, Dwarfism,hypergonadotropic hypogonadism.

INTRODUCTION

Turner syndrome being the inherited chromosomal aneuploidy condition presented with an incidence of 1 out of 2500 live female births .[2] The classical form is 45,X0 karyotype which is 60%,where the remaining 40% have a structural X chromosome abnormalities and an others can be some mosaic (45,x / 46,xx)-1/4th in some of the other patients. Henry Turner in 1938 described this condition and loss of X chromosome was identified in this syndrome in 1959 .Turner syndrome occurs due to nondisjunction of chromosome, loss of chromosome during gametogenesis resulting in 45,X0 Karyotype .[3,4]

2% of all zygotes - 45 ,X0 Karyotype

- 99% of 45,X conceptuses abort spontaneously
- 7%(1 in 15)of spontaneous abortuses – 45,x karyotype

The main features in early infancy is lymphedema(swelling in an arm or leg caused by a lymphatic system blockage) ,thickened nuchal fold ,low hairline ,cardiac anomalies.[5]In early childhood –unexplained Stunting of growth and characteristic features like Abnormal nails ,shield chest ,abnormal carrying angle ,recurrent ear infections are seen .Turner syndrome should be considered in all females with pubertal delay associated with hypergonadotropic hypogonadism , short stature in turner syndrome is sine qua non. So there is a loss of homeobox –containing gene on the pseudoautosomal region of p arms of X and Y chromosomes . [6]

The distinctive features of Turner syndrome is micrognathia resembling a fish mouth appearance ,high arched palate with poor dentition [2],epicanthal folds in the eyes ,ptosis in the eyelids ,low set ears ,webbed neck with low hairline ,recurrent otitis infection ,Broad,shield–like chest,poorly developed areolae ,skeletal defects –short 4 th metacarpals ,cubitus and genu valgum ,wrist deformity (made-lung) .cardiovascular anomalies : more on the left side of the heart –coarctation of the aorta (10%),aortic stenosis,bicuspid aortic aneurysm.[7]In kidneys, there are abnormal pelvicalyceal collecting system, abnormal positioning and vasculature of kidneys ,recurrent urinary tract infections are common ,[8] gastrointestinal symptoms (intestinal telangiectasias ,hemangiomatosis),autoimmune disease disease (hashimoto thyroiditis).metabolic cause like 50% impaired glucose tolerance .

Intrauterine growth restriction is common in all these patients with the average deficit in birth line of 2.6 cm (-1.24 SD).Height for age deficit occurs majorly in the first three years of age ,absence of pubertal growth spurt.[9]

CASE REPORT

A 14 years aged adolescent female was brought to the paediatric outpatient department in our hospital with the complaints of fever for 5 days , productive cough for 3 day ,stunting of growth in terms of height when compared to peer groups noticed in the recent past.No atopic history and history of chest deformity in family members. Examination revealed webbing of neck,low hairline (fig 2) ,shield chest(fig 1),nail dystrophy (fig 3),Tanner staging of breast and pubic hair development of the child is stage1 ,corresponding to pre pubertal stage. On systemic examination – cardiovascular system-systolic murmur of grade 1 is detected .her height measured 114 cm(less than 3rd percentile) ,weight was found to be 25 kgs (less than 3rd percentile).Body mass index was 15kg/m2 .

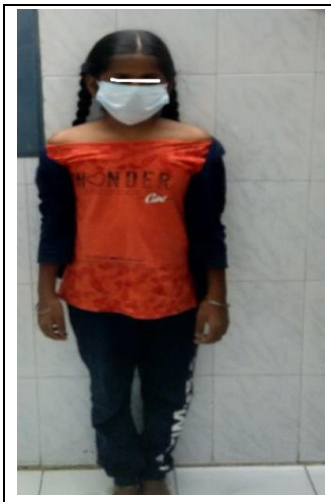

fig:1(SHIELD CHEST)

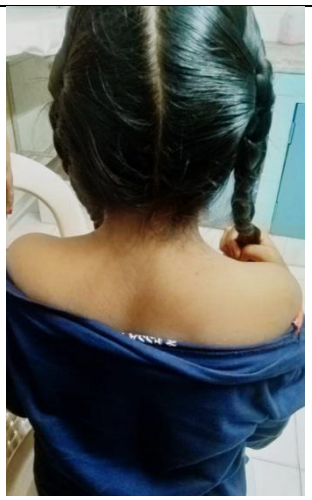

fig :2 (WEBBED NECK WITH LOW HAIR LINE)

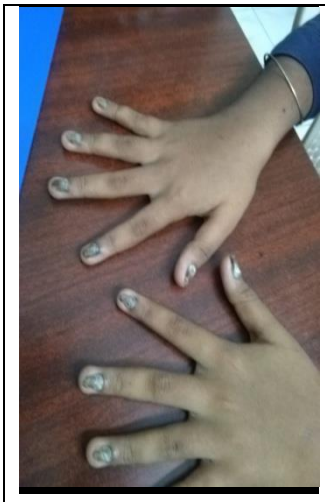

fig :3(NAIL DYSPLASIA)

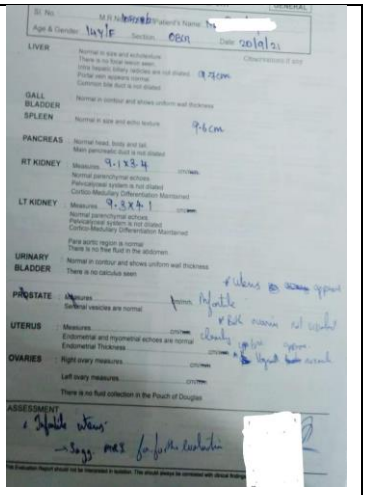

Fig:4 (USG ABDOMEN INFANTILE UTERUS)

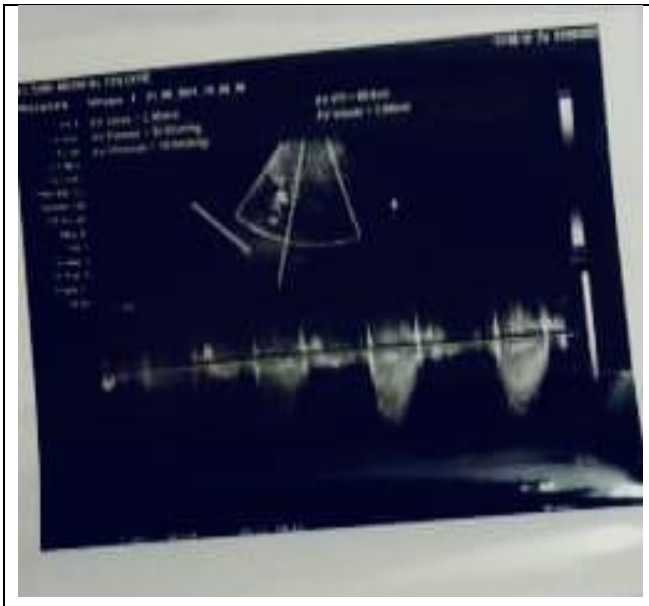

fig :5(BICUSPID AORTIC VALVE)

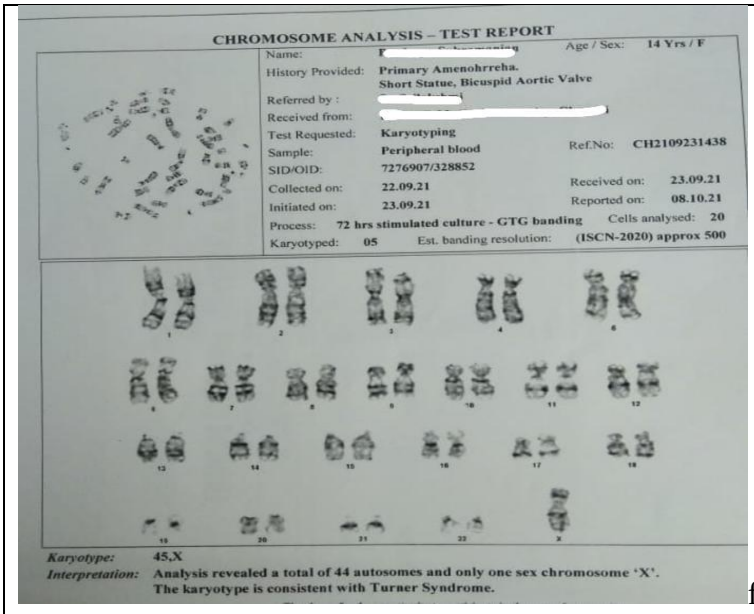

fig :6 (KARYOTYPE- 45,X)

INVESTIGATIONS :

1)FSH- **Raised** (20.2 m IU/ml), LH- **Raised** (2.32 m IU/ml) .

2)THYROID FUNCTION TEST- NORMAL

3)TESTOSTERONE LEVEL – NORMAL(0.16 ng/dl)

4)USG ABDOMEN SHOWED **INFANTILE UTERUS** ,BOTH OVARIES NOT VISUALISED

5)**ECHO FINDING REVEALED** Congenital heart disease ,situs solitus ,levocardia .AV-VA concordance ,NO PDA/COA/ASD/VSD/,**BICUSPID AORTIC VALVE** /THICKENED

VALVE ,MILD AORTIC STENOSIS (AV VELOCITY -2.85 m/s) AVPG -32 mmhg /AVMG-20 MMHG),TRIVIAL AORTIC REGURGITATION ,Normal cardiac chambers ,no RWMA at rest ,Good biventricular function(EF- 68%) FIG:5

6)CHROMOSOME ANALYSIS –(45,X0) ANALYSIS REVEALED A TOTAL OF 44 AUTOSOMES AND ONLY ONE SEX CHROMOSOME 'X'. THE KARYOTYPE IS CONGRUOUS WITH TURNER SYNDROME

On the basis of classical signs,laboratory investigations, karyotyping finding –**diagnosed a case of TURNER SYNDROME**

DISCUSSION

Gonadectomy is not mandated unless TYPY LOCUS in Y fragments are present .[10] There is a risk of gonadoblastoma in these patients.Oocyte cryopreservation done in case of turner mosaicism .Fertility preservation to be done in patients with persistent ovarian function .Ovarian cortical tissue preservation is feasible and requires surgery which in experimental phase.Routine fertility preservation in girls under 12 years of age however is not recommended .[11,12] Pregnancy complication is high due to cardiovascular defects and other complication

Further management of height – FDA approved GROWTH HORMONE is the treatment regime followed to attain adult height ,early initiation of growth hormone therapy will result in a favourable outcome. Modest additive effect are seen with low dose of estrogen.Age of initiation ,Dose duration,Number of injection per week ,Compliance are the factors influencing the treatment outcome .

The child was rendered symptomatic treatment and was referred to psychiatric counselling in the view to achieve social mingling ,self -contentment and acceptance of self - physicality.Gynecologist counselling and opinion was sought in terms of fertility management .

CONCLUSION:

Growth hormone therapy to attain appropriate height for age ,Hormonal substitutional therapy for feminisation ,Psychiatric counselling ,Family counselling and acceptance in the society is the mainstream treatment approach. Genetic counselling in parents is mandatory for subsequent pregnancy.[13,14]

Advancement in epigenetic analysis , knowledge of genetic and molecular technology will be a booming threshold for a better understanding and treatment of turner syndrome in near future .[15]

CONFLICT OF INTEREST:

Authors declare no conflict of interest

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