

## Case Report

# A Rare Case of Turner's Syndrome with Deaf-mutism Reporting at Government Medical College, Patiala, Punjab, India.

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### Abstract:

Abstract: Turner syndrome (TS), also known as Congenital ovarian hypoplasia syndrome, is a neurogenetic disorder characterized by partial or complete monosomy-X. Despite an often conspicuous phenotype, the diagnostic delay can be substantial and the average age at diagnosis is around 15 years of age. Morbidity and mortality are increased in women with Turner syndrome compared with the general population and the involvement of multiple organs through all stages of life necessitates a multidisciplinary approach to care. Treatment may include growth hormone therapy at appropriate time for short stature and estrogen therapy to help stimulate sexual development.

### Keywords:

Turner's syndrome, ovarian failure, lymphedema, deaf-mutism

## Introduction

Turner's syndrome (TS) is a rare disease with known genotypic and phenotypic variability that affects females in a ratio of 1:2,000 to 1:5,000 live births. This syndrome is caused by a partial or total deletion of sexual chromosome. Classical turner syndrome has complete missing of one X chromosome. About half of the subjects have a karyotype 45, X; 20%-30% have mosaicism, and the remainder have structural abnormalities<sup>1</sup>. The complete or partial loss of the X chromosome causes the phenotypic manifestations of Turner's syndrome, including: short stature, webbing of the neck, low posterior hairline, cubitus valgus, cardiac malformations<sup>2</sup> (e.g., coarctation of the aorta, aortic root dilatation), kidney malformations (e.g., horse shoe kidney), and ovarian dysgenesis<sup>3</sup>. There is high incidence of turner's syndrome, 3% of every conceived pregnancy but as 99% of them are spontaneously aborted, the reported incidence is very low.

| Genetics of turner's syndrome |          |
|-------------------------------|----------|
| 45,X                          | 50%      |
| 46,X,i(Xq)                    | 15%      |
| 45,X/46,XX mosaics            | 15%      |
| 45,X/46,X,i(Xq) mosaics       | about 5% |
| 45,X, other X abnormality     | about 5% |
| Other 45,X/? mosaics          | about 5% |

### Case report -

12 years old female reported to GMC, Rajindra Hospital Patiala, state Punjab, India with increasing swelling in the neck and difficulty in breathing. Child was second order female, born from non-consanguineous marriage. Maternal age at the time of conception was 20 years. On examination, HR- 88

bpm, BP – 126/78 mmhg (50th centile for age), low hairline, epicanthal eye folds, low set ears, high arched palate, webbed neck, short stature (<3rd centile), shield chest (broad chest with widely spaced nipples), cubitus valgus, scoliosis, lymphedema, pubic hair not developed, no menarche, mental retardation (IQ – 71) and neuro-cognitive disability were found. On investigations, Hb-11gm/dl, TLC 6700, N60L35, Platelets 4.8 lacs, PBF was microcytic-hypochromic with no polychromasia. RBS – 92, RFTs – Blood urea 17mg%, serum creatinine 0.6mg%, electrolytes: sodium-133meq/litre, potassium – 5.4meq/litre, calcium 7.9mg%. LFTs were normal. The child was found to have hypothyroidism, ovarian failure, X-ray hand showed short 4th and 5th metacarpals, on USG pelvis, uterus was not visualised and B/L small ovaries were seen. Hearing assessment was B/L refer. Karyotyping confirmed it as a case of

Turner's Syndrome.

Pictures showing phenotype of Turner Syndrome- webbed neck, shield chest, widely spaced nipples, cubitus valgus (A) epicanthal eye folds(B), low set ears and short webbed neck (C,D), scoliosis (E) and cubitus valgus and lymphedema (F)

The present case is deaf-mute which usually is due to recurrent otitis media. But in this child there was no such history. In this case the association of deaf-mutism may be due to narrow Eustachian tube.

### Discussion :

Turner syndrome is a rare condition characterized by female hypogonadotrophic hypogonadism, infertility, short stature, endocrine and metabolic disorders, an increased risk of autoimmune disease, as well as other medical conditions such as cardiovascular disease. The condition, which is associated with a completely or partially missing X chromosome, was first described 100 years ago by three independent clinicians, Seresewskij, Ullrich and Turner<sup>4</sup>. In a small percentage of cases of Turner's syndrome, some cells have one copy of X chromosome and the other cells have one copy of X chromosome and some Y chromosome material. These individuals develop biologically as girls but the presence of Y chromosome material increases the risk of developing a type of cancer called gonadoblastoma.

The association of otitis media, hearing loss and TS was reported in the early 60's, being confirmed by later studies<sup>5</sup>. It is recognized that individuals with TS have a higher incidence of middle ear disease and hearing problems than non-TS subjects. The associated hearing impairment has been described as both conductive and sensorineural and mixed indicating both middle and inner ear involvement due to abnormal eustachian tube structure and function<sup>6</sup>.

Recently some studies have described the role of the SHOX gene (short stature homeobox containing-gene) in the pathophysiology of the short stature and hearing problems in TS<sup>7</sup>. Parkin et al did a study in



turner syndrome patients where 91 % of subjects suffered from middle ear disease whilst the incidence of SNHL was 9%8. The pathophysiology of the sensorineural hearing loss in TS is not known, but theories regarding estrogen deficiency, the cell cycle delay hypothesis, IGF-1 deficiency and the possible role of the KDM6A gene can be highlighted9.

Following a diagnosis of Turner syndrome, management includes evaluation for other associated abnormalities like cardiac anomalies, renal anomalies, and learning disabilities. Screening should be a part of the baseline evaluation, and patients should undergo periodic screening thereafter. At initial diagnosis, patients should get renal ultrasonography and cardiovascular evaluation, including echocardiography in infants and children, and MRI in older girls and women.

#### Conclusion :

This is a rare case of Turner's syndrome with deaf-mutism presented at a pubertal age . Hormone replacement therapy was started at a low dose, 6mcgestradioldaily after measuring the baseline LH and FSH levels. Although the child had a height less than 3rd centile and was a candidate for GH replacement therapy, due to financial constraints it was not started. Child was also advised for hearing aid for her hearing disability10.

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