Pacific Journal of Medical and Health Sciences
ISSN: 2456-7450 | Vol.3, No.4, 2021, pp-33-37

PJMHS
PACIFIC JOURNAL OF MEDICAL AND HEALTH SCIENCES

Case Report

Recurrent Anaemia with Turner Syndrome: A Case Report

K.R. Sharma[1], Jagdish Vishnoi[2], Urvansh Mehta[3]*, Aseem Aasdhir[3], Harsh Patel[3]

[1]Professor and Head, [2]Professor, [3]PG Resident

Department of General Medicine, Pacific Medical College and Hospital, Udaipur, Rajasthan, India

*Corresponding author Email: urvanshmehta@live.com

ABSTRACT

Turner Syndrome is the most common genetic anomaly manifesting in females as primary ovarian failure, Infertility and short stature. There are numerous systemic manifestations reportedwith Turner Syndrome. Here we present a case of 19-Year-old female, diagnosed with Turner syndrome but with two uncommon features. Recurrent anaemia and jaundice.Genetic anomaly associated with Dimorphic anaemia and Hepatic derangement can be an isolated incidence here or an associative factor, further clinical research is required to derive an outcome.

KEYWORDS: Ovarian failure, Karyotyping, Dimorphic anaemia, Jaundice, Genetic anomaly.

INTRODUCTION

Turner Syndrome is the most common sex abnormality related to chromosome occurring in females. The incidence rate is around 1 in 2000-2500 live births[1].Turner syndrome is a genetic condition in which the affected females are missing an X Chromosome either completely or partially.[2] Before the discovery of karyotyping, Turner Syndrome was first reported in females with amenorrhea, webbing of neck, Joint deformities such as cubitus valgus and short stature. This was documented in a paper published by Henri Turner in 1938 from Oklahoma[3]. Turner Syndrome is also occasionally known as Ulrich syndrome because another physician Otto Ulrich had reported similar features in an 8-year-old female many years before Henri Turner's reporting.[4]

The most characteristic and classical symptoms seen consistently in females with Turner Syndrome is short stature followed by Primary Amenorrhoea. It can be manifested along with other structural anomalies such as webbed and short neck, shield like chest with wide spacing of nipples, short or absent fourth metatarsal/ metacarpal bone, high arched palate, low set ears etc.[5]

We report a case of 19-year-old Female with primary amenorrhoea, recurrent anaemia and short stature diagnosed with, Turner Syndrome.

CASE REPORT

A 19-year-old female, presented to the General Medicine OPD with complaints of

- Primary amenorrhoea
- Failure to gain height since 10 Years
- Easy fatiguability since 20 Days
- Decreased Appetite since20 Days
- Fever since 20 Days
- Yellowish discolouration of the eyes since 10 Days

Figure 1: Patient of Turner Syndrome

Arm Span	**124 cm**
Height	**129.5 cm (Father's height: 154.5 cm)**
Weight	**26 kg**
BMI	**16.1**
Upper Segment	**54 cm**
Lower Segment	**64 cm**
US/LS Ratio	**0.84**
Mid Upper Arm Circumference	**16 cm**

On physical examination and general appearance, the patient had short stature and severe pallor and Icterus (Fig. 1). Patient had consulted multiple doctors for the same complaint before the day of admission but had no relief. Patient never attained menarche and there was no visual development of secondary sexual characters. Patient had no sustained gain in height and weight after the age of 9 Years.

Patient gave a history of poorscholastic performance. On appearance patient had course facial features with soft dysmorphic signs. Any other associated anomalies such as Polydactyly, Goitre, colour blindness was absent.Patient had a wide spaced carrying angle (Cubitus Valgus) with syndromal short stature. Patient's secondary sexual characters were poorly developed. Patient was provisionally diagnosed the patient with Syndromal Short Stature?Turner Syndrome with Amenorrhoea and Anaemia with Jaundice.

Patient's peripheral blood film findings were as mentioned below:

- Sparsely distributed, microcytic, macrocytic and hypochromic RBC's with marked anisopoikilocytosis.
- Tear drop cells and pencil cells with Polychromasia
- Pancytopenia
- Morphology consistent with Dimorphic Anaemia

Her **haemoglobin was 3.2 gm%, but her serum iron was 233.7 µg/dl.** She had history of recurrent anaemia and blood transfusions for 2 times in the past also. Patient's **Follicle Stimulating Hormone (FSH) levels were 67.73 mIU/ml.** While her **Oestradiol level was <10 pg/ml (21-251 pg/ml).** We even ordered a **Testosterone level which was 10.6 ng/dl (10.83 -56.94 ng/dl).** Patient's thyroid function tests were within normal limits. This laboratory evaluation gave us a typical picture of **Hypogonadotropic Hypogonadism. Patient also had B12 and Vitamin D deficiency – 156.0 pg/ml (187-883 pg/ml) and 10.62 ng/ml (20-32 ng/ml) respectively.** Patient's Liver function tests were as follows;

- Toal Bilirubin: 2.98 mg/dl (0.2-1.0 mg/dl)
    - o Direct Bilirubin: 0.66 mg/dl (0.1-0.3 mg/dl)
    - o Indirect Bilirubin: 2.32 mg/dl (0.1-0.7 mg/dl)
- SGOT: 98 U/L (9-49 U/L)
- SGPT: 39 U/L (9-49 U/L)
- ALP: 46 U/L (38-126 U/L)
- Total Protein: 6.20 (6.4 – 8.4 g/dl)
    - o Albumin: 4.36 (3.5-5.0 g/dl)
    - o Globulin: 1.84 g/dl (2.5-4.0 g/dl)

Patient was further investigated radiologically. An ultrasound of whole abdomen revealed **hypoplastic small uterus (approx. 30 X 7 X 9 mm) with tiny hypoechoic ovary like structures in both adnexa (maybe a representative of streak gonads).** The Endometrium was indistinct from the adjacent myometrium (maybe due to extremely thin ET). Patient's 2D Echocardiography report was as below

- Bi-Cuspid aortic valve with eccentric closure line in M-Mode.
- Mild peak gradientacross the aortic valve of 12 mmHg. There was no Aortic Stenosis or coarctation of aorta. No aortic regurgitation.

An X Ray of left wristand elbow for bone age was done **(Fig. 2).** The X Ray reported **Fusion of epiphysis of lower end of the ulna (Under process)** and **Fusion of epiphysis of lower end of radius (Under process).Bone age fell between 14 – 17 Years.**

Ultimately, a blood karyotype analysis was done which showed a **Karyotype patter of 45, X.** The proband cytogenetically **abnormal** sex chromosome suggestive of **Turner Syndrome (Fig. 3.)**.

Patient was given 3 blood transfusions for Anaemia and Oestrogen supplements (lifelong) for the disorder. Oestrogen was given in form of oral tablets 0.625mg X Once Daily lifelong.

She responded well and her blood picture attained normal levels. She was discharged on Oestrogen supplements and advised regular follow ups.

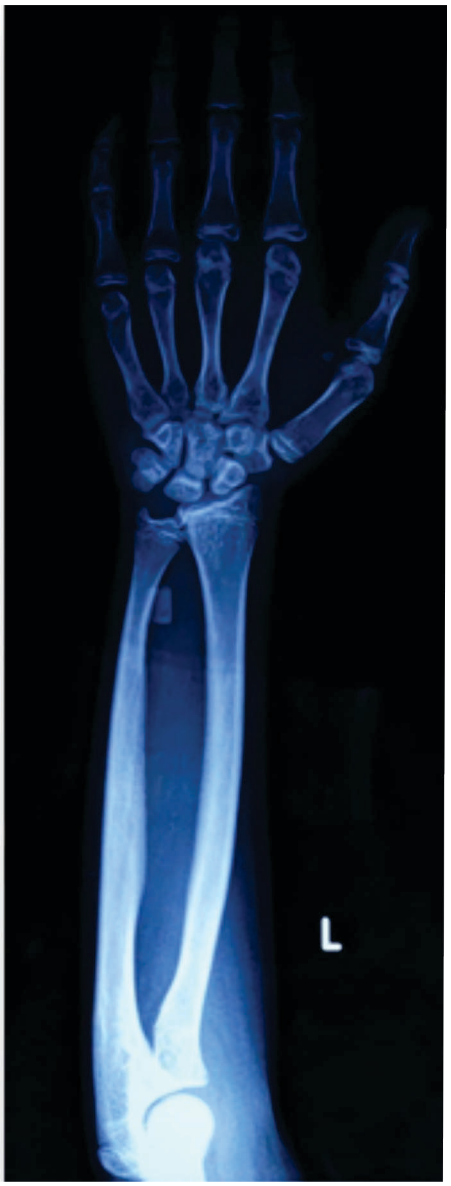

Figure 2: X – Ray Left Wrist and Elbow

Figure 3: Blood Karyotyping Report

DISCUSSION

Turner Syndrome, a common chromosome abnormality is a very important cause of short stature along with ovarian insufficiency in females.

The primary pathological basis for it is the Haploinsufficiency $\rightarrow$ 45,X (found in around 45% of Turner live births)[6]. Similarly other uncommon chromosomal anomalies reported with Turner syndrome includes **46,XX , 46,XY, del(p22.3), X-autosome translocation, unbalanced, del(q24) and idic(X)(q24) etc.**[7]

Around 95% of Turer Syndrome patients suffer from short stature. Although this patient's US/LS ratio was 0.84, an increased US/LS ratio is found in more than 90% of Turner syndrome patients. Cubitus Valgus and Genu Valgum is seen in around 40% of patient having a fairly common representation. Our patient also had an increased carrying angle (Cubitus Valgus). Given the underlying ovarian failure, 95% of turner syndrome patients suffer from primary amenorrhoea and infertility. Similar instance is seen in form of hypoplastic gonads, gonadal dysgenesis.[8,9,10]Several cardiovascular abnormalities are consistent with Turner Syndrome, with half of the patients presenting with elongation of transverse aortic arch and about one third of the patient with aortic valve abnormalities such as Bi-Cuspid aortic valve. Although rare but Coarctation of the aorta is also reported.[11,12,13]Renal and renovascular anomalies such as horseshoe kidney and collecting system anomalies are seen in around one third of the patients. The presence of thyroiditis is around 30% but with advancing age the incidence rate is increasing. About 70% of Turner Syndrome patient's present with recurrent otitis media and by adulthood about half of the patients develop hearing loss (more commonly sensorineural).[14]

Sparse clinical research and reporting is available showing presence of anaemia and jaundice with Turner Syndrome, but that has been attributed to gastrointestinal bleeding.[15,16,17]

CONCLUSION

Short stature, primary infertility and ovarian failure can lead to severe impact on a female's mental, social, and physical wellbeing. Societal pressure can also increase along with the poor scholastic performance If diagnosed early and timely regular follow up and treatment can give a patient suffering from Turner Syndrome a relatively normal life.

We also present an uncommon presentation here, in form of recurrent anaemia and jaundice. This calls for a newfound interest in research for Turner Syndrome research, maybe we are really missing another important clinical manifestation.

CONFLICTS OF INTEREST: None

FINANCIAL SUPPORT: None

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