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TURNERS SYNDROME MOSAIC VARIANT 45, X[10]/46,X,I(X)(Q10)[10] WITH HYPOTHYROIDISM– A CASE REPORT

General Medicine

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ABSTRACT

Turner syndrome (TS) was first described as a genetic disorder in 1938, and was found to result from the X chromosome being partially or completely absent. Turner Syndrome with 45, X karyotype was observed in 1-2 percent of human conceptions, 10 percent of pregnancy losses in the first trimester, and 1 percent of pregnancy losses are still births. More than 99 per cent of 45,X fetuses end up with abortion, typically through the 28th week of gestation, suggesting that living 45,X individuals must have mosaicism for another cell line (5,6)
We report a case of an 18-year-old Delhi resident presented with history of Amenorrhoea in the Department of General Medicine at SGT University Gurugram, poorly developed secondary sexual characters and increased forgetfulness. Her karyotyping revealed Mosaic variant 45, X[10]/46, X, i(X)(q10)[10], which is a very rare variant and therefore we are reporting this case.

KEYWORDS

INTRODUCTION

Turner Syndrome is among the most prevalent chromosomal abnormalities among female live births . The estimated frequency is 1/2500 (2). Primary amenorrhea, short stature and infertility are clinical findings. The physical characteristics are low nuchal hair line, small mandibula, low-set ears, valgus cubitus. The phenotypic characteristics also include nail hypoplasia, high palate, swelling of the hands and feetin the neonatal period, short fourth metacarpal bone, discrete nipples and wide thoracic cage. It is also possible to see cardiac and renal anomalies, hypothyroidism, hearing and vision disorders, gastrointestinal and dermatological problems, and neoplasms in TS (3).

Although due to the presence of dysmorphic findings some of the patients may be diagnosed at birth, the diagnosis is delayed until childhood, adolescence or later. Intelligence is not affected, in general. However, in 70 per cent of patients (4), learning difficulties that affect nonverbal, perceptual, motor and visuospatial skills can be seen. About 50 per cent of patients with TS have 45, X karyotype, and isochromosome Xq is the most common structural X chromosome abnormality.

A CASE REPORT

We have reporteda case of an 18-year-old Delhi resident with history of Amenorrhoea, poorly developed secondary sexual characters and increased forgetfulnessin the Department of General Medicine at SGT University Gurugram. She also had a history of breathlessness while walking a distance of up to 1 kilometer. She had poor performance in school and when she finally dropped out of school, she was studying in the seventh standard. She also had complaints of blood in vomitus and black coloured stools (two episodes each) for two days.

Past History: She has a history of taking some traditional Ayurvedic medication for beginning of menses . She had no history of hypertension, mellitus diabetes, tuberculosis, jaundice or any other chronic disease.

Family History:

Mother of the patient was obese and married at age 16. She had seven normal vaginal deliveries at home, the first and the second girls died at birth. The patient we report, is their family's third girl. Her younger sister is the fourth child and is alive. Her mother's fifth, sixth, seventh children, all died at birth. Her mother had an early menopause at the age of thirty and died at the age of forty because of a cardiovascular problem. Her father is also of a short stature. Her younger sister was twelve years old when she started with her menses and is having regular menstrual cycles. She is also short-statured.

Fig 1

A thorough general physical examination was done , and the findings were as follows. Her Blood pressure was 80/60 mmHg. At the time of admission her pulse rate was124 per minute. The height was 122 cm and she weighed 22 Kg. The BMI was estimated at 14.8 kg / m2.

Clinical physical examination showed broad spaced nipples, absence of secondary sexual characteristics, namely absence of axillary and pubic hair and absence of breast development, irregular development of external genitalia: labia majora and labia minora were not well developed. The Chest, Cardiovascular, Central Nervous System exam was normal.

On Abdominal Examination: She had approximately 5 cm of firm, smooth, non-tender hepatomegaly and 6 cm of splenomegaly. Figure 1 shows a photo of the girl.

LABORATORY INVESTIGATIONS

Her investigations revealed a Haemoglobin level of 6.6gm/dl, an RBC count of 2.97million/cm, MCV of 76.8fl, PCV of 22.8%, MCHC of 28.9L% and an MCH of 22.2pg.

Her Liver function tests revealed a Total Bilirubin of 1.4mg/dl, Direct Bilirubin-0.4mg/dl, AST- 63 IU/L,ALT-42 IU/L,ALP-171IU/L. Her Iron profile revealed a serum iron- 22.3, TIBC-424.7,%Transferrin saturation-5.25.

Her Thyroid Profile revealed a TSH of 14 micro IU/ml, a free T4 of 1mg/dl and a free T3 of 3.0 pg/ml. Her LH and FSH during the follicular phase were 25.8mIU/ml and 118.64mIU/ml, respectively.Her Cytogenetic report showed in figure 2, reveals 45,X[10]/46,X,i(X)(q10)[10] as her karyotype. Her ultrasound whole abdomen showed hepatosplenomegaly, a hypoechoic cystic area in

segment 7 of the right lobe and absent uterus and bilateral ovaries.

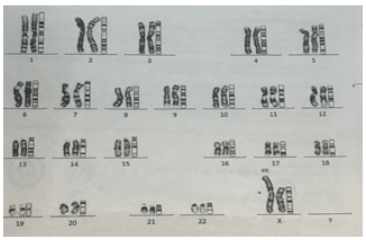

Figure 2

DISCUSSION

Homer et al. assessed differences between patients with sex chromosome mosaicism in clinical features and biological parameters. They reported that aneuploidy accounted for 8 per cent or more of a smaller height and early Menarche accounted for 10 per cent of aneuploidy.

However in a study by Hook et al., it was noted that the ratio of those with non-mosaic 45, X to those with mosaic 46, XX/45, X was 108 to 8 or about 13.5 to 1. in embryonic and fetal deaths The difference in the ratio of live births to embryonic and fetal deaths is significant at level 0.001. A case of Turner's syndrome mosaic variant is presented in this article. In this case, we report a 18-year-old female with Turner's syndrome variant isochromosome Xq.

Fertility and sexual development are often significant concerns in patients suffering from Turner syndrome. In a study by Hovata O et al. it is stated that the ovaries develop but typically degenerate in early childhood or during fetal life. Our case too presented with delayed development of secondary sexual characters.

However, in 2 to 5 per cent of patients with Turner syndrome, spontaneous menstruation and childbirth occur, which can be explained by substantial 46, XX/45, X mosaicism, with normal cell populations in the ovaries. Spontaneous puberty occurs in 5 to 10 percent of women with Turner's syndrome. Our patient had short stature and her height was 122 cm. These cases can have different clinical features. (8)

In our patient the family history was significant as her mother had seven normal vaginal deliveries out of which five children died at birth or following birth and she is the third girl child . Sutton Jerica et al. stated that Turner syndrome patients are likely to ask their family physicians about reproductive potential, and age-appropriate counseling on infertility treatments can significantly mitigate the diagnosis' adverse psychological impact. Because as spontaneous pregnancy is possible, if sexually active, patients should be advised on birth control. (9) In vitro fertilization (using harvested oocytes and cryopreserved before completion of the ovarian regression) is studied in young women with Turner syndrome. (10)

As we have already discussed that fertility and sexual development is a crucial part of this disorder, Bondy CA et al. studied the patients of Turner's syndrome for evaluation and treatment of this disorder. Spontaneous pregnancy carries substantial risks; therefore, preconception counselling has a great importance.Cardiac echocardiography or magnetic resonance imaging (MRI) play an important role. If pregnancy takes place, it is required to monitor the pregnancy as a high-risk obstetrics case.

In addition to reproductive counseling, management of cardiovascular risk factors such as hypertension, diabetes, hyperlipidemia should be done. calcium and vitamin D supplementation are also provided to these patients to prevent osteoporosis along with an ongoing sex hormone therapy. In addition our patient has hypothyroidism as she has TSH levels of 14 micro IU/ml and has been managed using Eltroxin.

At first adult visit, a baseline dual-energy x-ray absorptiometry scan is recommended for evaluating bone mineral density.In in 8 to 42 percent of patients over time, Aortic root dilatation is seen, Hence, Adult women should continue to receive high-quality aorta echocardiography or MRI every five to ten years to evaluate the need for surgical correction. For young girls and women the psychosocial impact of Turner syndrome can be substantial. These effects can be caused by infertility (in decreasing order of patient importance); short stature; and impaired development of sexual characteristics, most notably lacking libido. (9) Doctors should raise specific concerns from patients, address them individually and recommend a comprehensive psychoeducational assessment.

CONCLUSION

Isochromosome X[46, X, i(X)]: isochromosome is a structural chromosome aberration consisting of two short arms or two long arms, derived by division of centromere. X chromosome's most frequent structural abnormality is 46, X, i(Xq). In TS cases with or without mosaicism by Wolff DJ et al. (7), isochromosome frequency was stated to be 15-18 percent.

In our case, a female 18-year-old was diagnosed as a case of a mosaic variant of Turner's syndrome 45, X[10]/46, X, i(X)(q10)[10] with hypothyroidism.

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