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A Rare Case of Mosaic Turner Syndrome

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Abstract
Turner syndrome is a genetic disorder in which a female is partly or completely missing an X chromosome. Turner syndrome affects one in 2,000 to 5,000 female live births. Henry Turner first described the condition in 1938.In 1964, it was determined due to chromosomal abnormality. The chromosomal abnormality may be present in just some cells in which case it is known as Turner Syndrome with Mosaicism. In these cases, the symptoms are usually fewer or none occur at all. The diagnosis based on physical signs and genetic testing.

Keywords: *Turner syndrome, Genetic disorder, Mosaicism, X-chromosome, genetic testing, Female sex.*

Introduction

Mosaic Turner Syndrome is a rare genetic disorder with genotype 45x/46xx. When a sex chromosome is lost during early stages of embryonic development resulting in growth of some cells with single X-chromosome. Turner syndrome affects one in 2,000 to 5,000 female live births. Henry Turner first described the condition in 1938. In 1964, it was determined due to chromosomal abnormality. The chromosomal abnormality may be present in just some cells in which case it is known as Turner Syndome with Mosaicism. In these cases, the symptoms are usually fewer or none occur at all. The features of mosaic turner correlates with the relative percentage of 45X cells within the body compared to46xx cells

Case History

A 30 year old female patient presented with abdominal pain of 4days duration and exertional breathlessness 2 months duration. There is no history of cough, fever, burning micturition, vomiting or swelling of feet.

Past history of episodes of abdominal pain. History of recurrent episodes of lower respiratory tract infections since childhood. She is born out of non-consanguinous marriage .Other two siblings are normal. Menstual history of attained menarche at the age of 14 years. Having regular menstrual cycles with normal flow since menarche. On general examination-Patient is conscious, coherent, with short stature, short neck, hypertelorism, high arched palate, crowding teeth, low posterior hair line, scoliosis, hypoplastic 4th and 5th fingers and pallor. Height-130 cms, Weight-38 kg, vitals- normal.

Systemic examination of CVS -Pan systolic murmur of grade 3/6 present in lower left sternal boarder, P2 loud in pulmonary area, Respiratory system-Normal vesicular breath sounds, no adventitious sounds.
Investigations-CBP-Hb:7 gms,TC-6300 cells /mm^3, DC- P 62%, L 33%, E-5%, ESR-20 mm/ 1^{st} hr ,Platelet count-adequate. RFT-Serum creatine-0.6 mg/ dl, Blood urea- 26 mg / dl .RBS-94 mg / dl. 2D-ECHO- Large VSD with bidirectional shunt, severe PAH, Dilated RA/RV, good LV function. Ultra Sound Abdomen And Pelvis- Right renal caliculi, Normal ovaries, Normal uterus. Harmonal Assays-Thyroid Profile-TSH-2.8 µiu/lit, T4-11pmolLit ,T3-1.8 nmol/Lit. S. Alkaline Phosphatase-11IU/Lit.S. Calcium-10.5mg/dl,S. Intact Parathyroid Harmone-42.8 pg/ml.S. Growth Harmone- 10.8 ng/ml. Chromosomal Analysis-Abnormal results revealed the presence of two cell lines.13 metaphases showed the presence of a single X-chromosome where as 17 metaphases showed normal female karyotype. MTS45X[13]46XX[17].

Figure:1-crowded teeth and short neck.

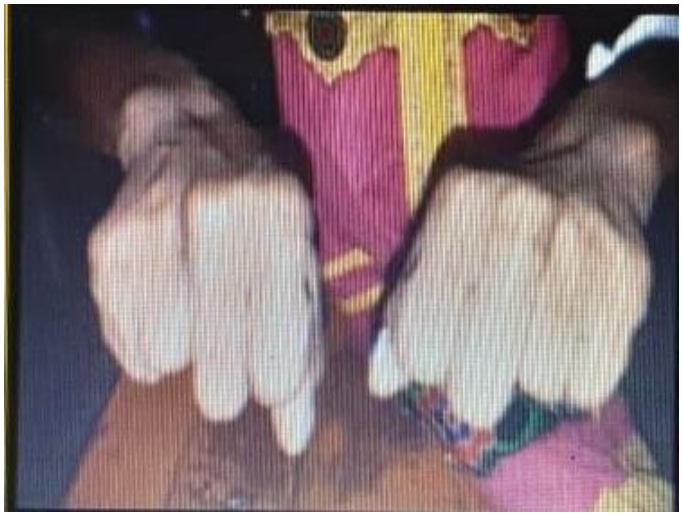

Figure: 2-Hypoplastic 4 th and 5^{th} fingers.

Discussion

Turner syndrome is associated with multiple chromosome abnormalities including 45,X and 45,X/46,XX and 45,X/47,XXX and 45,X/46,XY. Mosaic turner is a rare type of turner syndrome. Turner syndrome with 45, X/46, XX low-level mosaicism may not be detected on standard karyotype and FISH analysis of larger numbers of cells can be useful for diagnosis. The American College of Medical Genetics (ACMG) provides guidelines for karyotyping procedure specific to Turner syndrome. The College recommends karyotyping a minimum of 30 cells due to the high incidence of mosaicism, unless mosaicism is encountered within the first 20 cells. When there is a high clinical suspicion of Turner syndrome in a patient with a 46, XX karyotype, cytogenetic study of a second tissue type (such as skin biopsy for cell culture or buccal smear for FISH) is advised. In the above case the examination features and musculoskeletal abnormalities were in favour of genetic syndrome probably turners syndrome but the normal menstrual history and normal reproductive organs on ultrasound raised the suspicion of mosaic turner syndrome .so karyotyping done and the results are in favour of 45X/46XX mosaic turner syndrome.

Conclusion

Turner syndrome occurs in about 1 in 2000 to 1 in 5000 female births. Among them 60% may be

mosaic turner with variable clinical features correlating with relative percentage of 45x cells in the body compared to 46XX, cells in the body compared to 46 xx cells.

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