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Mosaic Variant Turners Syndrome A Rare Case Report

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Abstract : A 6 year old girl was brought to us with complaints of being short. On examination she was a case of proportionate short stature. There were no syndromic or dysmorphic facies. Endocrine evaluations were normal as also were the cardiology and ultrasound findings. Karyotyping was done which was reported as Mosaic Turners Syndrome. This case is being presented for Mosaic Turners Syndrome presenting as isolated short stature.

Keyword : Mosaic Turners Syndrome, Short Stature Mosaic Variant Turner's Syndrome -A Rare Case Report

INTRODUCTION

Turner syndrome is caused by a missing or incomplete X chromosome. The genes affected are involved in growth and sexual development, which is why girls with the disorder are shorter than normal and have abnormal sexual characteristics. The incidence of Turner's Syndrome is 1 in 1500- 2500 live born females. Mosaicism occurs in a higher proportion than that seen with any other aneuploid state.

CASE REPORT

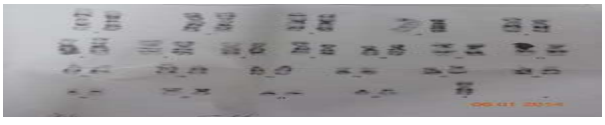
A 6 year old developmentally normal girl was brought by her mother with complaints of the child being short when compared to other children of her age. She was first of the two siblings and her sister was younger to her by 3 years. She belonged to Class 3 socioeconomic status. She lived with her parents and sister. The per capita income of the family was Rs.3000 and there was no social conflict in the family No webbing of neck and no facial dysmorphism

Bone Age-2 years

Her height was 90 cm which was below the third percentile and her weight was 10 kg which was also below the third percentile for age. Both were based on the CDC charts. The upper segment to lower segment ratio was 1.14 which is normal for a 6 year old. The mean parental height of her parents was 155 cm. Her head circumference was normal for her age. She was thus a case of proportionate short stature. Her Tanner staging was 1 with no breast bud and no pubic hair.Her cardiac status was normal which was confirmed by e;ectrogram, chest radiography and echography.There were no other visible anamolies.

As a first line of investigation complete haemogram was done which did not reveal any infection. Serum calcium, phosphate and serum alkaline phosphatase were within normal limits as also was her random blood sugar. This was followed by assessment of bone age which was 2 years. Her opthalmological and otorhinolaryngology evaluation was normal.Ultrasound abdomen was also normal. Other first line investigations were normal and so we proceeded with second line investigations. The values of free triidothyronine, free thyroxine and thyroid stimulating hormone were 3.2pg/ml, 1.8ng/ml and 4.629 mIU/ml respectively. These were within normal limits. Growth Hormone stimulation assay was done following administration of clonidine 0.15 mg/m2 per orally with regular monitoring of blood pressure. The basal and post stimulation assays at 30, 60 and 90 minutes were normal. Meanwhile the karyotyping was reported as mos 46, X, +mar (21)/45, X (9) which was consistent with a diagnosis of Mosaic Variant Turner's Syndrome. There were two cell lines. One cell line had loss of one X chromosome while the other cell line had loss of one X chromosome along with a marker chromosome of indeterminate origin. Karyotyping showing 46 chromosomes with one absent X chromosome and a small marker chromosome (mar)

Karyotyping showing 45 chromosomes with one absent X chromosome



Following the diagnosis the parents were counseled and karyotyping of the other sibling was also done which was normal. Tissue transglutaminase levels were also measured as there is a higher incidence of celiac disease in Turner's Syndrome. It was found to be normal. The serum level of Follicle stimulating Hormone was elevated. The girl is on regular follow up with us and

hormone replacement therapy with estrogen has been planned for her at the age of 12-13 years in consultation with the endocrinologist and gynaecologist along with regular monitoring of thyroid function test and tissue transglutaminase. This case is being presented for the isolated manifestation of mosaic variant of Turner's Syndrome as short stature.

DISCUSSION

Turner's Syndrome was first described by Ullrich in 1938 and is also termed Ullrich-Turner Syndrome. It is the only hyper gonadotrophic hypogonadism in which there are prepubertal clinical manifestations. Turner syndrome encompasses several conditions in females of which monosomy X is most common. It is a chromosomal abnormality in which all or part of one of the sex chromosomes is absent or has other abnormalities. In some cases, the chromosome is missing in some cells but not others, a condition referred to as mosaicism. Turner syndrome is caused by nondisjunction. Women with Turner syndrome are usually sterile and cannot have children but mosaic variants can bear children. In about 20 percent of cases, one X chromosome is abnormal. It may be shaped like a ring, or missing some genetic material. About 30 percent of girls with the disorder are only missing the X chromosome in some of their cells. This mixed chromosome pattern is known as mosaicism. Girls with this pattern may have fewer symptoms because they still have some normal cells. In the parents of a Turner syndrome the risk of recurrence is not increased for subsequent pregnancies. Rare exceptions may include the presence of a balanced translocation of the X chromosome in a parent, or where the mother has XO mosaicism restricted to her germ cells.

One of the missing genes on the X chromosome is the SHOX gene, which is responsible for long bone growth. So the girls who have the disorder are unusually short. Other missing genes regulate ovarian development, which influences sexual characteristics. Turner syndrome patients appear to have a stocky appearance, arms that turn out slightly at the elbow, a receding lower jaw, a short webbed neck, and low hairline at the back of the neck, lymphedema, short stature, broad chest and widely spaced nipples, low hairline, low-set ears, sterility, rudimentary ovaries gonadal streak, amenorrhoea, obesity, shortened fourth metacarpal, small fingernails, webbed neck from cystic hygroma in infancy, aortic valve stenosis, horseshoe kidney, visual impairments, ear infections and hearing loss, high waist-to-hip ratio, attention deficit hyperactivity disorder and nonverbal learning disability. Other features may include micrognathia, cubitus valgus, soft upturned nails, palmar crease, and drooping eyelids. Less common are pigmented moles and a high-arch palate.Any of the features may b present or absent depending on the mosaicism. The average height of an untreated woman with Turner syndrome is 144-150cm.

Cardiovascular manifestations include coarctation of aorta, bicuspid aortic valve, hypoplastic left heart, partial anomalous venous drainage, aortic dilatation, dissection and rupture and increased risk of bacterial endocarditis. About half of the cases are diagnosed within the first few months of life by the characteristic physical symptoms. Other patients are diagnosed in adolescence because they fail to grow normally or go through puberty. Turner syndrome may be diagnosed during pregnancy with a chorionic villus sampling or amniocentesis. Usually, fetuses with Turner syndrome can be identified by abnormal ultrasound findings (i.e., heart defect, kidney abnormality, cystic hygroma, ascites). Although the recurrence risk is not increased, genetic counseling is often recommended for families who have had a pregnancy or child with Turner syndrome. Despite the excellent postnatal prognosis, 99% of Turner-syndrome conceptions are thought to end in spontaneous abortion or stillbirth.

As it is a chromosomal condition there is no cure for Turne r's Syndrome. Replacement therapy with estrogens is indicated but there is no consensus as to at what age it has to be started. Many girls achieve heights greater than 150 cm with early initiation of treatment. The starting dose of growth hormone is 0.375 mg/kg/ week. Serum levels of IGF -1 should be monitored if the patient is receiving high doses of growth hormone. Oxandrolone has also been used in combination with growth hormone to treat short stature. The cost of these two drugs is forbidding.

Successful pregnancies have been carried to term with ovum donation and in vitro fertilization. Girls with few signs of puberty can have ovaries with follicles. Psychosocial support for the girls is an important component of treatment. With early and appropriate medical care and ongoing support, most patients can lead normal, healthy, and productive lives.

CONCLUSION

This case is being presented for the rarity of presentation of mosaic variant Turner's Syndrome with isolated short stature alone without any other manifestations.So it is necessary to think of Turner's Syndrome mosaicism when a girl child presents with short stature alone without any other manifestations.

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