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Case Report

Turner syndrome: a neglected case.

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Abstract:

An 18 years old girl hailing from Mirpur-10, Dhaka was admitted into Paediatrics Dept. of Delta Medical College & Hospital, Dhaka on 4th March 2010 with the complaints of delayed sexual development, bilateral ear discharge with hearing problem for 2 years, occasional headache and some behavioral abnormality for the last 1 month. She is the second issue of nonconsanguinous parents. Her other sibs were healthy. She was clinically diagnosed as a case of Turner syndrome and confirmed by karyotyping. She was given symptomatic management as because hormonal treatment would not be beneficial at this age.

Introduction:

Turner syndrome is a numerical chromosomal disorder affecting females. It occurs in 1 of 2000 to 1 of 5000 live born female.[1,2] Turner syndrome is characterized by complete or partial monosomy of X chromosome and defined by a combination of phenotypic features, the most consistent with short stature and gonadal dysgenesis. The syndrome is named after Henry Turner, an American endocrinologist who described the phenotypic feature of the syndrome and was the first to initiate oestrogen replacement therapy.[3] The syndrome is confirmed by karyotyping. Affected girls are usually treated with recombinant growth hormone alone or in combination with oxandrolone, oestrogen replacement therapy. Psychological support is an integral part of the treatment.[4] Modern reproductive technology such as in vitro fertilization using donated ova is used to help woman with turner syndrome for reproduction.[5]

Case Report:

An 18 years old girl hailing from Mirpur-10, Dhaka was admitted into Paediatrics Dept. of Delta Medical College & Hospital, Dhaka on 4th March 2010 with the complaints of delayed sexual maturation, bilateral ear discharge with hearing problem for 2 years, occasional headache and some behavioral abnormality for 1 month. She is the second issue of nonconsanguinous parents. Her other sibs were healthy.

She was working as a maidservant in a family.

On examination she had some behavioral abnormality. Such as anxiety, depression and fear. She was short with height 131cm, arm span 142 cm, upper segment: lower segment= 0.8. Her height was below the lower limit of midparental height. She had shield shaped chest with widely spaced nipple. She had wide carrying angle. She was hypertensive (blood pressure was 140/100 mm of Hg). She had short 4th metacarpal bones of both hands, short 4th metatarsal bones of both feet and short 3rd metatatersal bone on left side. Her pubertal staging (sexual maturity rating) shows- stage3 breast development, there was no pubic or axillary hair, vulva was well developed. There was no organomegaly.

Lab investigations: Karyotyping was 45,X0. Audiometry showed bilateral mixed type hearing loss. Complete blood count, Echocardiogram and thyroid function showed normal findings. Ultrasonography of the whole abdomen showed hypoplastic uterus and absent ovaries.

She was diagnosed as turner syndrome associated with hypertension and behavioral problem.

The patient as well as her parents were properly ccunseled. She was treated with beta blocker, multivitamins, tranquilizer and psychotherapy. No hormonal treatment was given as the case was late in diagnosis and her uterus not developed properly. The role of hormonal therapy in such case is doubtful. After improving the general condition she was discharged on request. She was advised for follow up but she did not come for follow up later on.

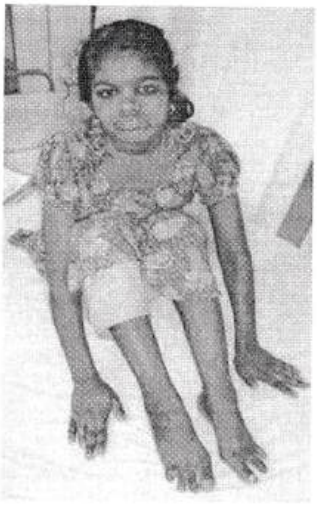

Fig. 1: A patient with Turner syndrome

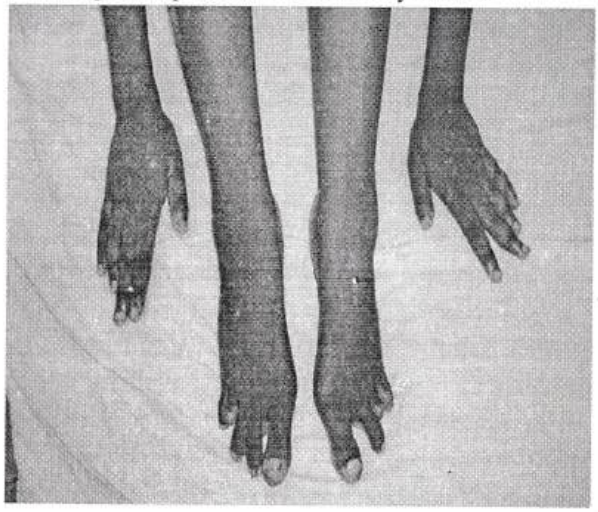

Fig. 2: Features of bony dysplasia in both hands &feet.

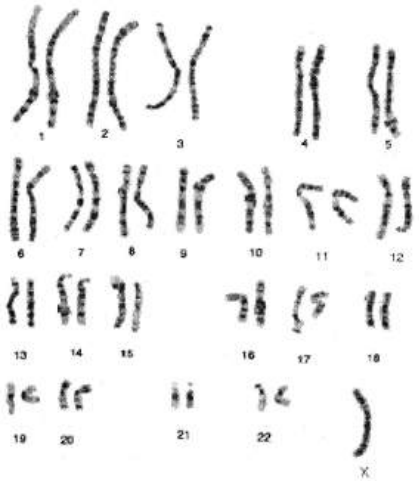

Figure 3: Karyotyping of Turner syndrome

Discussion:

Turner syndrome is the combination of characteristic phenotypic features accompanied by complete or partial absence of the second X chromosome with or without mosaicism. Fifty percent of patient have 45,X chromosomal complement. Rest 50% patients have mosaic pattern including 45,X/46,XX; 45,X/46,X,i(Xq); 45,X/46,X,r(X);45,X/46fra.In about two third of women with Turner syndrome , the normal X chromosome is maternal origin.[6-8] Monosomy of X results from nondisjunction as a result of failure of sex chromatids to separate during meiosis in the paternal gamete or in the early embryonic division. Monosomy X is the only chromosomal monosomy compatible with life. The second X chromosome in 46,XX female is inactivated forming the Barr body.[9] Fifteen to twenty percent of X chromosome gene on the "inactive" genes are transcribed and are termed pseudo-autosomal that contains short stature homeobox (SHOX) gene, which is only specific gene known to contribute to the turner syndrome phenotype. A study in NIH (national institute of health study) of united state between 2001 and 2005 showed the following phenotypic features of turner syndrome-short stature 98%, ovarian failure 92%, sensorineural hearing loss 55%, hypertension3o%, impaired glucose tolerance 45%, neck webbing 33%, skeletal anomalies 10%, renal anomalies 30%, liver enzyme deranged 38%, obesity 36%.[10]

Karyotyping which shows complete or partial deletion of one of the X chromosome is the basis of definitive diagnosis. Antenatal diagnosis can be done by antenatal screening based on ultrasound findings (such as increased nuchal translucency, cystic hygroma, coarctation of aorta, renal anomalies or polyhydramnios), abnormal triple or quadriple serum screening. The diagnosis is confirmed by amniocentasis or chorionic villous sampling. In a study of 19 European registries, 67.2% of prenatally diagnosed cases were detected by anomalies on ultrasound (69.1% had one anomaly, 30.9% had two or more anomalies.).[11] Mosaic karyotypes were more associated with positive maternal serum and less likely had an associated ultrasound anomaly. Postnatally a karyotype should be considered to rule Turner syndrome in all phenotypic female. Elevated circulatory follicle stimulating hormone during infancy and adolescence indicate gonadal failure and points to Turner syndrome. To detect associated problems some investigations should be done. Echocardiogram and magnetic resonance angiography are done to detect cardiac problem, ultrasonography of abdomen to detect kidney problem, thyroid function test and thyroid antibodies should

be checked periodically, audiological assessment should also be done.

A multidisciplinary approach is necessary to improve the quality of life of patient with Turner syndrome. Treatment of patient with this syndrome should focus primarily on the associated clinical manifestations. Therapeutic strategies include surgical treatment of associated malformations (such as cardiac anomalies), oestrogen replacement therapy, somatropin (growth hormone), oxandrolone (anabolic steroid) supplementations and therapeutic approaches. Somatropin is a biosynthetic form of growth hormone that is used in the treatment of short stature including Turner syndrome since 1985.[12,13] Better response depends on early initiation of somatropin therapy.[14,15] Somatropin therapy has also beneficial effect on blood pressure, voice/speech changes and body proportions in patient with turner syndrome.[16-18] Somatropin therapy shows considerable variability when anabolic steroid and oestrogen therapy is used concomitantly. Oestrogen replacement therapy should be used to induce the secondary sexual characteristics. As oestrogen therapy decrease the response to somatropin it should be timed as to prevent any negative effect on growth while inducing puberty at an appropriate age (at 11-12years).[19,20] Several recent studies have suggested that conventional pubertal induction does not produce optimal development of the uterus in Turner syndrome.[21-24] It is not clear from these studies whether impaired uterine development was due to delayed oestrogen therapy, too low dose or due to use of synthetic androgenic progestin.

Conclusion:

Early diagnosis and multidisciplinary treatment at appropriate time can improve growth and development and the quality of life of a female with Turner syndrome. She can enjoy a normal family as well as fertile life.

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