CASE REPORT

Turner Syndrome

Ravinder K. Gupta, Ritu Gupta*, Sunil Dutt Sharma**

Abstract

Turner Syndrome is one of the important chromosomal disorders characterised by loss (total or part) of sex chromosome. The manifestations being peripheral edema, short stature, extra skin fold, webbing of neck, renal and cardiovascular anomalies, sexual infantilism, learning disability etc. We present here a one month female baby who had classical features of Turner Syndrome. The karyotape analysis was consistent with the diagnosis.

Key words

Turner Syndrome, Chromosome, Peripheral edema

Introduction

Turner Syndrome is characterized by sexual infantilism, webbed neck, short stature, peripheral edema, lymphedema, renal and cardiovascular anomalies, gonadal dysplasia, some learning disability etc. (1-4). The condition is defined as the combination of the characteristic phenotypic features accompanied by complete or partial absence of the second X chromosome with or without mosaicism. This is most common monosomies in live born humans. The frequency at birth is 1/4000 to 1/8000 live births, but it occurs much more frequently in spontaneous abortions (1-4). We present here a female child who classical features of Turner Syndrome.

Case Report

One month female, first in order was noticed to have swelling of both hands and feet since birth. The baby was passing urine adequately. There was no history of fever, cough, refusal of feed, rash or any abnormal movements. The baby was first in order and was born of non-consanguineous marriage. The marriage has taken place almost a year back and age of mother at the time of birth was 25 years. Antenatal period was uneventful. This full term baby was delivered pervaginally at home and the birth process was uneventful. There was no significant family history of such complaints. On examination, the baby was active, afebrile, with good cry and had normal physiological neonatal reflexes. The vitals were maintained. Her weight was 2300 gms. length was 46 cms. and head circumference 34 cms. There was no pallor, jaundice or cyanosis. There was pitting edema over dorsum of hands and feets and this edema was not present on rest of the body. The baby had webbed and short neck and had low posterior hair line. The chest appeared broad and had widely spaced nipples (both the nipples were ouside midclavicular line). The cardiovascular, respiratory and abdominal examination was clinically normal. The baby underwent investi-gations. The hemoglobin was 17gm/dl, TLC-7500/mm3, polymorhs-53, lymphocytes-45 and monocytes-2. Routine urine and 24 hours urine for proteins were within normal limits. RFT and LFT were also within normal limits. VDRL of mother and baby were negative. TORCH test was normal. X-ray chest and skull did not reveal anything abnormal. The ultrasound of abdomen was normal except that ovaries could not be localized. Echocardiography was normal. The baby was subjected to karyotype analysis. Every well spread metaphase plate contained 45 chromosomes after short-term lymphocyte culture. The karyotype prepared from metaphase plates showed XO condition.

In view of clinical details and laboratory finding the diagnosis of Turner Syndrome was made. The baby was managed symptomatically and followed.

From the Departments of Pediatrics, Acharya Shri Chander College of Medical Sciences and Hospital, Sidhra, Jammu, *Avad Pediatrics Clinic & **Health Services, Jammu (J&K), India.

Correspondence to : Dr. Ravinder K Gupta, 'The Nest' 209-A, Gandhi Nagar, Jammu (J&K), India

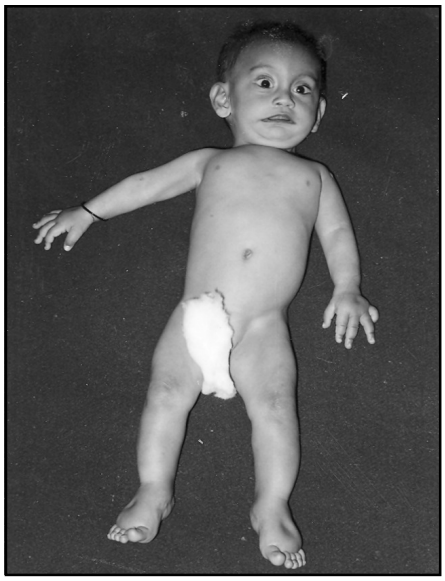

Fig. 1-a. Child with Turner Syndrome

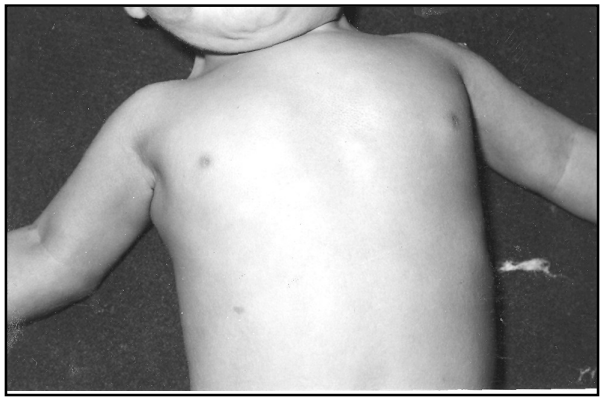

Fig. 1-b. Showing wide spaced nipples

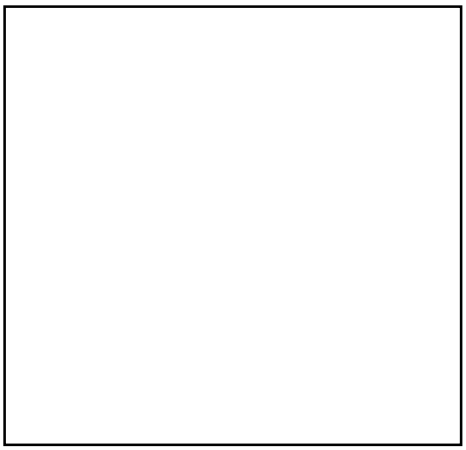

Fig. 1-c. Showing pitting edema dorsum of feet

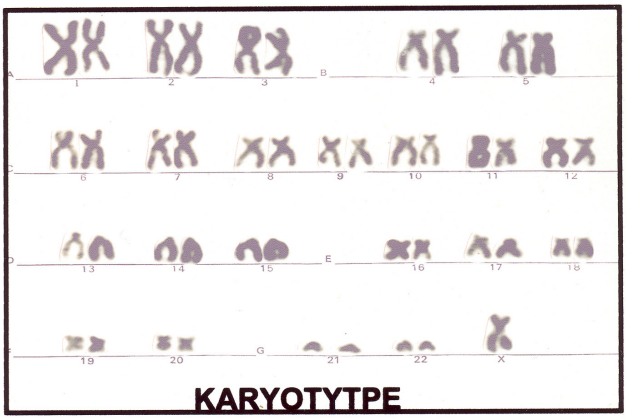

Fig. 2. Showing only one X chromosome, Other sex chromosome missing - X0

Discussion

Turner described a syndrome consisting of sexual infantilism, webbed neck, and cubitus valgus in adult females. Ulrich described an 8 years old girl with short stature and many of the some phenotypic features. The term Ulrich-Turner Syndrome is frequently used in Europe but rarely used in the United States (1). Many patients with turner syndrome are recognisable at birth because of a characteristic edema of the dorsa of the hands are feet and loose skinfolds at the nape of the neck. Low birth weight and decreased length are common (1-5). Our child was having edema of the dorsa of the hands and feet since birth and birth weight at birth was reported to be 2300 grams. Clinical manifestations in childhood include webbing of neck, a low posterior hair line, small mandible, prominent ears, epicanthal folds, high arched palate, a broad chest presenting the illusion of widely spaced nipples, cubitus valgus and hyperconvex nails (1-4). The case reported in this study had almost all the features as shown in figures (1a-1c & 2). Associated defects are common. Non stenotic bicuspid aortic valves in one third to one half of the patients are seen. In later life bicuspid aortic valve disease can progress to dilation of the aortic root. Less frequent defects include aortic coarctation (20%), aortic stenosis, mitral valve prolapse and anomalous venous drainage (1, 6). In a study of 170/393 females with Turner syndrome in Denmark, 38% of patients with 45 X chromosome had cardiovascular malformations compared with 11% of those with mosaic monosomy X: the most common were aortic valve abnormalities and aortic coarctation (6). Cardiac

evaluation in our child did not reveal any thing abnormal. One fourth to one-third patients have renal malformation on ultrasonographic examination. The more serious defects include pelvic kidney, horse shoe kidney, double collecting system, complete absence of one of the kidney and ureteropelvic junction obstruction (1). A significant decrease in percentage of detectable ovaries from infancy to later childhood was found (1). In our case on ultrasonographic examination of abdomen, no renal defect was detected, but ovaries could not be localised.

Recurrent bilateral otitis media develops in about 75% of patients. Sensorineurol hearing deficits are common and the frequency increases with age (1). Problems with gross and fine motor-sensory integration, failure to walk before 15 months of age and early language dysfunction often raise questions about developmental delay but intelligence is normal in most cases.

Half the patients with Turner Syndrome have a 45 X chromosome compliment. About 15% of patient are mosaics for 45 X, and a normal cell line (45, X/46, XX). Other mosaics with iso-chromosomes with rings, or with fragment, occur less often. The single X is of maternal origin in 50-70% of 45, X patients. The mechanism of chromosome loss is unknown and the risk for the syndrome does not increase with maternal age. The frequency of the 45, X karyotype at conception is about 3.0% but 99% of these are spontaneously aborted accounting for 5-10% abortuses. (1,2).

Our baby was also subjected to karyotype analysis. Every well spread metaphase plate contained 45 chromosome after short-term lymphocyte culture. The karyotype prepared from metaphase plates showed XO condition (as shown in figure 3 & 4). The most common skeletal abnormalities are shortening of the 4th metatarsal and metacarpal bones, epiphyseal dysgenesis in the joints of the knees & elbows, madelung deformity, scoliosis and in older patients, inadequate osseous mineralization. However in our case there were no such findings.

Recombinant human growth harmone replacement therapy with estrogen and psychosocial support are the treatment modalities (1,6). Prenatal chromosome analysis for advanced maternal age has revealed a frequency of 45, X/46, XX that is 10 times higher than when diagnosed postnatally. Our child was managed symptomatically. Unfortunately the child's follow up was lost.

Conclusion

A strong possibility of Turner Syndrome should be kept in mind if a child presents with edema of hands and feet since birth.

References

1. Zinn AR, Page DC, Fisher EMC. Turner Syndrome: The Case of the missing X chromosome. *Trends Genet* 1993 ; 9 : 90.
2. Gicquel C, Gaston V, Cabrol S *et al*. Assessment of Turner Syndrome by molecular analysis of the X chromosome in growth retarded girls. *J Clin Endocrinal Metab* 1998 ; 83 : 1472.
3. Ross JL, Roeltgen D, Feuillan P *et al.* Use of estrogen in young girls with Turner Syndrome. *Neurology* 2000 ; 54 : 164-70.
4. Sybert VP. Cardiovascular malformations and complications in Turner Syndrome. *Pediatrics* 1998 ; 101 : 11.
5. Rapaport R. Disorders of the Gonads. In Behrman RE, Kiliegman RM, Jenson HB (eds). Nelson Textbooks of Pediatrics. 17th edn. WB Saunders Company, Philadelphia 2004 ; pp 1931-34.
6. Gupte S, Frank S. Genetics in Pediatric Practice. In : Gupte S (ed). The Short Textbook of Pediatrics 10th edn. Jay Pee Brothers, New Delhi. 2004 ; pp 576.