

## Case Report

# Turner syndrome in a neonate

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### ABSTRACT

Turner's syndrome (TS) is the common chromosomal abnormality. However, it is diagnosed rarely in the neonatal period. In many cases the diagnosis of TS may be delayed until childhood, when evaluation for short stature yields the diagnosis, or adolescence, when combination of growth failure and pubertal delay suggests the possibility of TS. Girls with TS are usually treated with growth hormone and oestrogen replacement therapies for short stature and oestrogen deficiency. A multidisciplinary team is usually required for management. Authors report an 11 day old infant who was diagnosed as Turner's syndrome. The classic clinical feature, lymphoedema clinched the diagnosis in our case which was confirmed by chromosomal analysis. The infant was discharged and asked to follow up for regular growth monitoring and parents were counselled regarding the condition.

**Keywords:** Lymphoedema, Turner's syndrome, Webbing of neck

### INTRODUCTION

Turner syndrome (TS) is caused by complete or partial absence of one of the X sex chromosome. The incidence is 1/2500 to 1/5000 live births. 20% are diagnosed at birth and rest during childhood or puberty.<sup>1</sup> Ninety nine percent of TS conceptuses abort spontaneously; TS accounts for ten percent of all first trimester miscarriages.<sup>2</sup> TS is characterised by webbed neck, short stature, widely set nipples and amenorrhea.<sup>3</sup> Authors report an infant who was diagnosed as TS and provide detailed clinical information, diagnosis and treatment.

### CASE REPORT

An 11 day old full term female infant born to IUFD1P1L1 mother via caesarean section i/v/o foetal distress, cried immediately after birth, presented with oedema of feet and over dorsum of hands since birth and decreased urine output since three days. There was no

history of feeding difficulty, lethargy and fever. Mother was registered and immunised. Mother's antenatal scans were suggestive of lymphedema and severe oligohydramnios (Amniotic fluid index: 4). No family history of similar disorders or other genetic diseases. On examination vitals were normal. Anthropometry: Weight: 2596 grams (50<sup>th</sup> to 90<sup>th</sup> percentile), Head circumference: 34cm (50<sup>th</sup> to 90<sup>th</sup> percentile); Length: 50cm (Upper segment: 30cm, Lower segment: 20cm), Chest circumference: 30cm.

Baby had non pitting edema till legs and over dorsum of hands (Figure 1 and 2), short neck, widely spaced nipples and low hairline (Figure 4). Urine output was adequate. Routine investigations were normal. Renal function tests were normal. Ultrasound abdomen was suggestive of horseshoe kidneys. 2D Echo revealed 2mm patent ductus arteriosus. Urine culture was suggestive of *Acinetobacter* sensitive to Ampitum, hence was started on Ampitum for 10 days. Repeat urine culture was no growth. Thyroid

function tests were normal. Follicle stimulating hormone (FSH) and Luteinizing hormone (LH) levels were high. Chromosomal analysis showed a karyotype of 45 XO (Figure 3). Parents were counselled regarding the condition and were advised DMSA Scan (Dimercaptosuccinic acid) and to follow up 3 monthly for growth monitoring.



Figure 1: Edema on the dorsum of feet.



Figure 2: Edema on the dorsum of the hands.

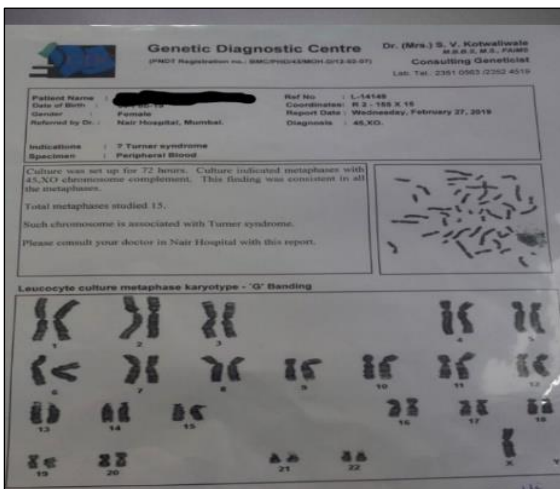


Figure 3: Karyotyping report.



Figure 4: Low hairline.

### DISCUSSION

TS is named since the first report of Turner in 1938.<sup>4</sup> TS is one of the monosomy syndrome in which affected individual can survive. Those patients who have mosaicism have presence of two or most distinct cell lines. The classical karyotype account for only 50% of the cases.<sup>5</sup> Short stature of TS phenotype is associated with homobox gene (SHOX), present on X and Y chromosome. The reduction in final height, changes in bone morphology and other features is due to haploinsufficiency of SHOX.<sup>5</sup> Those who have 45X/46XY mosaicism can be associated with male pseudohermaphroditism.<sup>5</sup> Patients during the neonatal period are usually 45XO, present as mosaics later in life.<sup>6</sup>

Turner syndrome is characterised by webbed neck, triangular facies, short stature, wide set nipples, amenorrhea, and absence of secondary sex characteristics.<sup>3</sup> Clinical manifestations vary and maybe subtle. Some affected girls, particularly those with mosaicism have only short stature and amenorrhea, without dysmorphism. Majority of patients with TS experience early ovarian failure.<sup>7</sup>

One third of patients with TS have cardiovascular anomalies; 75% of these have coarctation of aorta or a bicuspid aortic valve with increased risk of aortic dissection. Echocardiography and cardiac MRI are preferred imaging modalities for surveillance. There may be increased mortality in neonatal period if associated with hypoplastic left heart syndrome.<sup>8</sup> Renal anomalies occur in one third to one half of the girls which include pelvic kidney, horse shoe kidney, double collecting system, complete absence of one of the kidney and ureteropelvic junction obstruction. Renal anomalies may cause a predisposition to urinary tract infections. This patient had horseshoe kidney with urinary tract infections.

In utero, the ovaries have a decreased number of primordial follicles, these appear to undergo premature apoptosis and are usually absent by adult life. FSH levels

are usually elevated indicating early ovarian failure. Growth velocity decreases as early as 18 months of age. There is lack of spontaneous pubertal development from ovarian sex hormone insufficiency. The mean final height in girls with TS is 20cm below the mean of normal girls.<sup>9</sup>

TS is associated with various neurocognitive and psychosocial defects. Girls typically have normal intelligence. They may have difficulty with nonverbal, social and psychomotor skills. Characteristic neurocognitive and psychosocial profile are associated with TS. There are deficits in visual-spatial abilities and perceptual abilities.<sup>10</sup> Autoimmune conditions like thyroiditis, diabetes mellitus and celiac disease are associated with TS.<sup>11</sup> Other potential complications include infertility, strabismus, sensorineural hearing loss, orthodontic anomalies, skeletal abnormalities, learning difficulties and liver dysfunction.<sup>4</sup>

The diagnosis can be confirmed by careful examination of genetic material, usually in a blood sample. The diagnosis should be considered in a female foetus presenting with hydrops, increased nuchal translucency, cystic hygroma or lymphedema.<sup>12</sup>

Physicians play an important role in coordinating multidisciplinary management and in directly managing risk factors and complications because of the complexity of the disease. Treatment of TS includes monitoring and treating congenital heart disease and the use of recombinant growth hormone to tackle short stature. Growth hormone therapy is started as early as 12 to 24 months of age. Oestrogen replacement therapy is started at preteen years to promote the development of sex organs and to maintain bone density (usually initiation of oestrogen replacement before 13).

Growth hormone therapy is discontinued when a patient reaches a bone age of 14 years. Sex hormone therapy is generally continued throughout life.<sup>13</sup> 2-5% of TS patients of typical karyotype and those of 46 XX/45X mosaicism may experience spontaneous menstruation and pregnancy. The psychosocial impact may be significant for young girls and women due to infertility, short stature, and impaired development of sexual characteristics.

## CONCLUSION

It is essential to diagnose Turner's syndrome early so that appropriate treatment including hormonal therapy can be started to achieve normal growth and to induce puberty. Regular check-ups and proper monitoring can help most women lead a relatively healthy, independent life.

Management of TS during adulthood mainly involves the maintenance of reproductive health and associated cardiovascular diseases. A full understanding of this disorder and effective communication with patients and their family are crucial to improve the life quality of TS sufferers.

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## REFERENCES

1. Gicquel C, Gaston V, Cabrol S, Le Bouc Y. Assessment of Turner's syndrome by molecular analysis of the X chromosome in growth-retarded girls. *J Clin Endocrinol Metab.* 1998;83(5):1472-6.
2. Jones KL, Smith DW. *Smith's Recognizable Patterns of Human Malformation* 6<sup>th</sup> ed. Philadelphia: Elsevier Saunders; 2006;76-81.
3. Elias ER, Tsai AC, Manchester DK. *Turner Syndrome. Current Diagnosis and Treatment Pediatrics* 2009. 19<sup>th</sup> ed. New Delhi: Mc Graw Hill; 1002-1003.
4. Turner HH. A syndrome of infantilism, congenital webbed neck, and cubitus valgus. *Endocrinol.* 1983;2:566-74.
5. Gravholt CH. Clinical practise in Turner syndrome. *Nat Clin Pract Endocrinol Metab.* 2005;1(1):41-52.
6. Nandgaonkar BN, Shanbag VS, Gupta A, Varthkavi P, Udani RH. Turner syndrome in a neonate. *Indian Pediatr.* 1991;(28)812-4.
7. Hanton L, Axelrod L, Bakalov V. The importance of estrogenreplacement in young women with Turner syndrome. *J Women's Health.* 2003;12(10):971-7.
8. Natowicz M, Kelley RI. Association of Turner syndrome with hypoplastic left heart syndrome. *Am J Dis Child.* Feb 1987;141(2):218-20.
9. García-Rudaz C, Martínez AS, Heinrich JJ, Lejarraga H, Keselman A, Laspiur M, et al. Growth of Argentinian girls with Turner syndrome. *Ann Human Biol.* 1995 Jan 1;22(6):533-44.
10. Ross JL, Zinn A, McCauley E. Neurodevelopment and psychosocial aspects of Turner Syndrome. *Ment Retard Dev Disabil Res Rev.* 2000;6:135-41.
11. Bonamico M, Pasquino AM, Mariani P, Danesi HM, Culasso F, Mazzanti L, et al. Prevalance and clinical picture of celiac disease in Turner syndrome. *J Clin Endocrinol Metab* 2002; 87(12):5495-8.
12. Cassidy SB, Allanson JE. *Management of Genetic Syndromes.* 2<sup>nd</sup> ed. NJ John Wiley and sons, Hoboken. 2005.
13. Rappaport R. *Turner Syndrome. Nelson Text Book of Pediatrics* 2007. 18<sup>th</sup> ed. Philadelphia: Saunders Elsevier; 2386-2389.

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