

# Turner Syndrome; A Case Report With Growth Failure

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Turner syndrome which afflicts approximately 50 per 100000 females is characterized by retarded growth, gonadal dysgenesis, and infertility. Growth failure is a consistent finding at birth in infants with Turner's syndrome. Turner syndrome was diagnosed in a two-month-old female infant with intrauterine growth retardation. She had thriving problems, atypical appearance, and coarctation of the aorta. Her karyotype was 45,X, classic for Turner syndrome. With this case, we emphasized the necessity to investigate female infants born with low birthweight and growth failure during the first year of life.

**Key words:** Turner syndrome, growth, growth failure, intrauterine growth retardation

## Introduction

Turner's syndrome is a relatively common disorder occurring in 1 in 2000 live female births (1). Moderate intrauterine growth retardation results in birthweight and length below the mean in Turner's syndrome (2). Growth failure is a consistent finding at birth in infants with Turner's syndrome. However, the time of onset and pattern of growth deficiency is unknown (3). This disease can be detected before birth through chromosome analyses on cells from amniotic fluid (4).

X monosomy is the most commonly occurring sex chromosome anomaly, but the vast majority of Turner conceptuses are spontaneously aborted (1). We reported this fact to emphasize the necessity to investigate and to be careful with female infants born with low birthweight and having thriving problems during the first year of life for Turner syndrome.

## Case Report

A two-month-old female patient was admitted to our hospital with weight loss and failure to thrive.

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She was born weighing 1750 g at term with spontaneous vaginal delivery. Her birth length is not known. She lived in the incubator for two days. Although she was fed with breast milk for two weeks, thereafter she could not get it (Thereupon, the breast milk was taken by breast pumps and given to her for one week). After the third week, she was fed with formula milk which was given without diluting in suitable amounts. The infant's mother had used 4-5 analgesics due to tooth-ache during the pregnancy and she had had high fever for 3 days in the 5<sup>th</sup> month, but did not receive any drugs. During her pregnancy, she had a weight gain of 15-16 kg and also she was not seen by an obstetrician.

The baby was born of nonconsanguineous parents. The mother's first five-year-old child was healthy at birth whereas the second pregnancy resulted in an abortion. Our patient was the last child.

There was a problem in gaining weight, with loss of the turgor of a skin with a loose subcutaneous fat. She had micrognathia, a high-arched palate and prominent ears (Fig. 1). On physical examination, her height and weight were below the third percentile (44 cm and 2100 g respectively).

Hypochromic microcytic anemia was found in our patient. Liver and renal function tests, electrolytes, and blood glucose were all normal. Abdominal



Fig. 1. The patient with Turner syndrome.

ultrasonography was normal. In pelvic ultrasonography, although a uterus was found, ovaries were not seen.

Plasma levels of gonadotropins, particularly FSH were markedly elevated above those of age-matched controls during infancy (FSH: 5.2 mIU/ml, LH: 4.1 mIU/ml, and estradiol: < 20 pg/ml). Thyroid functions tests were found normal.

Two-dimensional color-Doppler, M Mode echocardiographic examination revealed dimensions and wall thickness of right and left ventricle appropriate for her age. It was observed that lumen of aorta in the origin of the left subclavian artery just in distal was narrower as to her age in suprasternal notch sagittal view the distal of stenosis. Maximum pressure gradient was 25 mm Hg continuous wave Doppler made (Fig. 2).

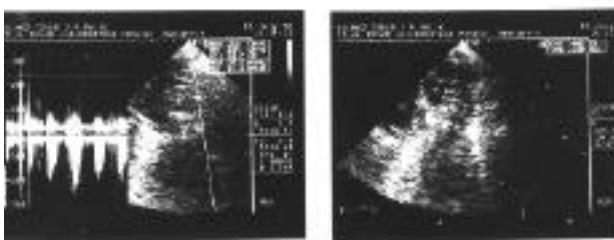


Fig. 2. Echocardiographic findings show coarctation of aorta

A peripheral blood culture technique modified in our cytogenetic laboratory was used in order to prepare the preparations. Giemsa-Trypsin-Giemsa band technique was used for the preparation. Twenty five metaphase plates were investigated microscopically and the chromosome analysis of the patient was diagnosed as 45,X (Fig. 3).

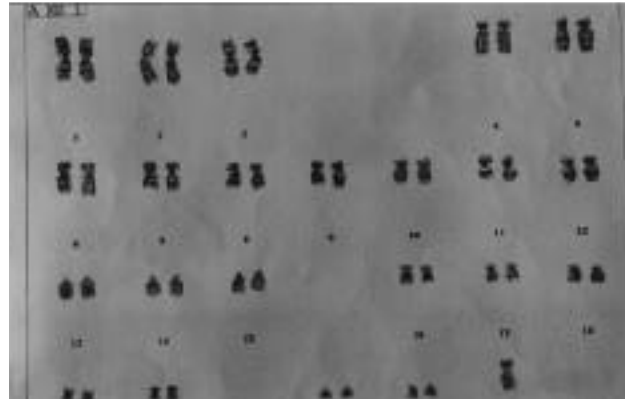


Fig. 3. A 45,X karyotype

### Discussion

In Turner syndrome, the most commonly observed karyotype is 45,X, followed by 45,X/46,XX mosaicism. Patients with 45,X karyotype are diagnosed at a younger age with mean adult height than those with 45,X/46,XX mosaicism (5). In another study, the highest somatic scores and the most severe clinical manifestation were noted in cases of pure 45X, Turner syndrome (6). 45,X embryos and fetuses are lost much more frequently (95%) (7). Our patient had the typical somatic features and the karyotype was 45,X in the examined cells.

Cardiac disorders especially aortic coarctation, should be mentioned. Echocardiography shortly after birth will show if there is any heart disorder. Twenty six % of Turner syndrome cases have congenital heart disease, whereas this is 2-3 % in the general population (8). In another study, cardiovascular anomalies are found in 45 % of patients with Turner syndrome. The most frequently associated cardiac anomalies are coarctation of the aorta and bicuspid aortic valve. A high absolute prevalence of bicuspid aortic valve (17.5 %) and aortic coarctation were observed relative to comparable series (9). Hypertrophic cardiomyopathy has been reported only in one case with this syndrome, but it is frequent in Noonan syndrome (10). A 23 -year-old female with Turner syndrome with a horse-shoe kidney had coarctation of the aorta with the thoracic aortic aneurysm (11). The only cardiac anomaly we determined in our patient is the coarctation of the aorta.

In one study, left renal agenesis and right hydro-nephrosis due to functional ureteropelvic junction obstruction was determined in an infant with Turner

Syndrome (12). This condition emphasizes the importance of routine imaging for early identification of potentially serious renal anomalies in patients being evaluated for short stature and possible Turner syndrome. Renal anomalies were not found in our patient.

In an investigation it is shown that by measuring the footlength and six long bones, fetuses with 45,X/46,XX mosaic Turner syndrome may demonstrate a lesser degree of growth retardation at least for footlength than those with a 45X, karyotype. In this study, they showed that growth failure beginning early in gestation is well-established by mid-pregnancy (3). In our patient with 45X, karyotype the growth retardation probably started in the early periods of pregnancy and became more evident in late pregnancy.

In one study, all cases with a positive fetal screening test or a positive screening obtained before an amniocentesis for other reasons were included. Serum marker levels and ultrasonographic findings were reviewed. The multiple-marker screening test appeared to detect Turner syndrome, as well as trisomies 21 and 18 (13). In our patient, if the periodical ultrasonographic examinations had been made, the growth retardation would have been established and the multiple marker tests should have been done during pregnancy. Moreover, the abortion in the second pregnancy of the mother is an important factor in the consideration of multiple marker tests. If our case had been seen by an obstetrician during her pregnancy the fetuses's growth retardation could have been determined by ultrasound and if her mother's abortion in her previous pregnancy could have been taken into consideration the multiple marker tests could have been done.

Temporary thriving problems during the first year of life should be mentioned. These problems usually disappear during the second year of life (14). The most outstanding finding in our patient was the problem of growth. The majority of infants with Turner syndrome have feeding problems. The reason for feeding problems, are oral anomalies, high arched palate, hypotonia in mandibular muscles, disfunctional tongue movements and disfunctional chewing skills. Their meal-times are significantly shorter than those of healthy infants (15). Our patient also had feeding problems and she had a high arched palate.

Genetic counselling must be given to parents with a suspicion of Turner syndrome in pregnancy. When counselling parents of a fetus with Turner's syndrome, they should be encouraged to ask questions and should be given sufficient time to ask these questions as often as they wish. There was an agreement that the frequency of prospective Turner parents induced abortion among was high in Denmark and most probably would be reduced if all parents were given prenatal counselling (14). We agree that more information about Turner's syndrome in general is needed in all countries.

In conclusion, genetic counselling should be given to parents with suspicion of Turner syndrome with intrauterine growth retardation during pregnancy with the help of ultrasonography and multiple marker tests. Also in female infants with intrauterine growth retardation and a problem of weight gain during the first months of life an investigation should be performed for Turner syndrome.

## References

1. Hook EB, Warburton D. The distribution of chromosomal genotypes associated with Turner's syndrome: livebirth prevalence rates and evidence for diminished fetal mortality and severity in genotypes associated with structural X abnormality or mosaicism. *Hum Genet* **62**: 24-27, 1983.
2. Neely EK. Turner Syndrome in Pediatric Endocrinology. Lifshitz F, Narcel Dekker (ed) INC, Newyork, 1996: 267-280.
3. Fitz Simmons J, Fantal A, Shepard TH. Growth parameters in mid-trimester fetal Turner syndrome. *Early Hum Dev* **38(2)**: 121-129.
4. Koeberl DD, McGillivray B, Seybert VP. Prenatal diagnosis of 45X/46XX mosaicism and 45X implications for postnatal outcome. *Am J Hum Genet* **57(3)**: 661-666, 1995.
5. Suri M, Kabra M, Jain U, Sanders V, Saxena R, Shukla A, Singh GV, Verma IC. A clinical and cytogenetic study of Turner syndrome. *Indian-Pediatr* **32(4)**: 433-442, 1995.
6. Temtamy SA, Ghali I, Salam MA, Hussein FH, Ezz EH, Salah N. Karyotype/phenotype correlation in females with short stature. *Clin Genet* **41(3)**: 147-151, 1992.
7. Kelly TE, Ferfuson JE, Golden W. Survival of fetuses with 45,X: an instructive case and an hypothesis. *Am J Med Gene* **42(6)**: 825-826, 1992.
8. Gøtzsche CO, B.Krang-Olsen, Nielsen J, Sorensen KE, Kristensen BO. Prevalence of cardiovascular malformations and association with karyotypes in Turner's syndrome. *Arch Dis Child*. **71**: 433-436, 1994.
9. Dawson-Talk KL, Wright AM, Bakker B, Piflick PT, Wilson DM, Rosenfeld RG. Cardiovascular evaluation in Turner syndrome utility of MR imaging. *Australas Radiol* **36(3)**: 204-209, 1992.

10. Conte MR, Bonfiglio G, Orzan F, Mangardi L, Camaschella C, Alfarano A, Brusca A. Hypertrophic obstructive cardiomyopathy in a patient with Turner Syndrome. *Cardiologia* **40(12)**: 947-949, 1995.
11. Kusaba E, Umada T, Iwakuma A, Nonaka K. Aortic aneurysm complicated coarctation of the aorta and Turner syndrome. *Kyobu-Geka* **48(13)**: 1115-1117, 1995.
12. Benjamin B, al-Harbi N. Renal agenesis and ureteropelvic junction obstruction in an infant with Turner Syndrome. *Ann Trop Pediatr* **17(1)**: 101-103, 1997.
13. Wenstrom KD, Williamson RA, Grant SS. Detection of fetal Turner syndrome with multiple-marker screening. *Am J Obstet Gynecol* **170(2)**: 570-573, 1994.
14. Konradsen B, Nielsen J, Sorensen G. First European Turner contact group meeting. *Copenhagen* **10-12**: 15-24, 1996.
15. Mathisen B, Reilly S, Skuse D. Oral motor disfunction and feeding disorder of infants with Turner syndrome. *Dev Med Child Neurol* **34(2)**: 141-149, 1992.