



Case Report

Turner Syndrome and Apparent Absent of Uterus : A Case Report

Munira Ferdousi¹

Abstract

A 15 years young fair girl presented with primary amenorrhoea. Findings on her physical examination included short stature, cubitus valgus, slightly webbed neck, absent of secondary sexual characters, broad chest with widely spaced nipples and absence of pubic & axillary hair. Except for the above, her systemic examination was unremarkable. A gynecological examination failed to reveal any abnormality. Ultrasound examination of abdomen revealed absent uterus and ovaries. Serum T3, T4 were within normal level, but TSH level was in slightly raised. Plasma FSH & LH were high with low estradiol level suggestive of hypergonadotrophic hypogonadism. Karyotyping confirmed the diagnosis of Turner's syndrome (45, X0).

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Introduction

Turner's syndrome, a disorder of females is characterized by the absence of all or part of a normal second sex chromosome, leading to a constellation of physical findings that often includes congenital lymphedema, short stature and a gonadal dysgenesis.^{1,2}

Turner's syndrome occurs in 1 in 2500 to 1 in 3000 live born girls. Approximately half have monosomy (45,X) and 5-10 percent have a duplication (isochromosome) of the long arm of one X (46,X,1(Xq)). Most of the rest have mosaicism for 45, X, with one or more additional cell lineages.

Historically, in 1938 Turner's described the syndrome which has become synonymous with his name. Currently the syndrome is referred to by such other designations as ovarian dwarfism, ovarian agenesis, syndrome of rudimentary ovaries and Bonnevie-Ullrich syndrome. We also currently use the term gonadal dysgenesis agree with Grumbach et al.³ and his associates introduced the gonadal dysgenesis.

The classic picture of turner's syndrome or gonadal dysgenesis is well known and is associated with a variety of other anomalies.⁴ In a review of the literature, the women involved are described as having primary amenorrhoea, are of short stature but not dwarfed and possess rudimentary ovaries. The sexual hair is scant or absent and there is conspicuous lack of breast development. The uterus and vagina are infantile in type. There is also an associated high urinary gonadotropin level & lowered urinary 17 ketosteroids. Many of these females have deep set ears, cubitus valgus, an increased carrying angle between the forearm and upper arm, and various ocular abnormalities. In the severe forms of the syndrome there are various degrees of webbing of the neck, coarctation of the aorta associated with an unexplained hypertension, deformity of the nails & a high arched palate.⁵

Following turner's report researchers have shown by investigations of the chromosomal sex patterns as determined by blood pressure⁶, skin biopsy⁷ and oral mucosal smears⁸ that the majority of patients with ovarian dysgenesis are chromatin negative.

¹ Assistant Professor, Department of Obs & Gynae, Rajshahi Medical College, Rajshahi.

Case history

A 15 years fair young girl from Mirpur, Kushtia, presented with the complaint of primary amenorrhoea, no breast development, no pubic hair like her younger & older sisters. Her height was 57 inches and weight 45kg. Her IQ level was well & good. Her past history is otherwise not remarkable. Detailed menstrual history revealed that she never had a menarche or vaginal spotting. There was no history of cyclical abdominal pain. Patient did not report any history of chemotherapy, radiotherapy, trauma or surgery to gonads. Past medical history was negative for mumps, tuberculosis or any major systemic illness.

Patient was a product of non-consanguineous marriage. She was born to a full term pregnancy with normal vaginal delivery at home. There was no history of obstructed labor, ante-partum or post partum haemorrhage, congenital birth defects, birth asphyxia or neonatal jaundice.



Fig. 1



Fig. 2

On close inquiry, it was revealed that she was considerably shorter than her peers. The patient reported that she started lagging behind in her height as she approached 8th or 9th year of life. She also noticed her secondary sexual characters failed to develop properly as she reached her pubertal age. She has normal intelligence.



Fig. 3

On examination, the patient was hemodynamically stable. She had a blood pressure of 110/80mm of

Hg. Her pulse was 75/min & regular and respiratory rate of 15/min.

Physical examination on hospitalization revealed a well nourished, reserved appearing female whose head was cove red with moderate growth of dark hair. Her height was.... & weight....Cubitus valgus was apparent. As reported by the ophthalmologist her eye ground, visual field and intraocular tension were normal. Skin turgor was good and the neck appeared proportionate to her body size. No webbing was apparent. The chest was the broad shield type (Lisser's sign). The nipples were widely spaced and she had absolutely no breast development. There was no axillary, pubic & labial hair growths.

Pelvic examination under general anaesthesia, the labia appeared undeveloped & immature. The external urethral meatus was normally placed and the clitoris presented as a small projection measuring 0.5mm in length. The vaginal outlet was virginal in type. On per rectal examination no cervix, no uterus, tubes or ovaries could be palpated.

Laboratory investigations

Routine laboratory studies revealed the blood count, total cholesterol, blood sugar and urine analysis to be within normal limit. There was no abnormality is detected in chest X-ray, ECG and Echocardiogram. Ultrasound of urogenital system showed no abnormalities in kidney and or pelvis but uterus and ovaries were absent. Her TSH was 6.98mIU/L. Her FSH level is 36.50 IU/L & LH level is 52.15 IU/L. and Estradiol level is 16.21 pg/ml.

Table : 1

Estimated Value		Normal Range	
TSH	6.98 mIU/L		0.3-5.0 mIU/L
LH	52.15 IU/L	Follicular	2.84-13.74 IU/L
		Ovulation	15.05 – 72.20 IU/L
		Luteal	1.72- 13.26 IU/L
FSH	36.50 IU/L	Menopausal	21.61 – 57.60 IU/L
		Follicular	1.82 – 7.75 IU/L
		Ovulation	5.17 - 29.25 IU/L
Estradiol (E2)	16.21 pg/ml	Luteal	0.75 – 4.15 IU/L
		Menopausal	28.21 -82.07 IU/L
		Female	31- 417 pg/ml
		Adult	5 – 63 pg/ml
		Post Menopausal	Upto 30000 pg/ml
		Pregnancy	

Chromosome analysis report

Karyotype: (Analysis performed by peripheral blood lymphocyte culture with G-banded)

Total no of chromosome: 45

No of autosome: 22

No of sex chromosome: X

Karyotype: 45, X

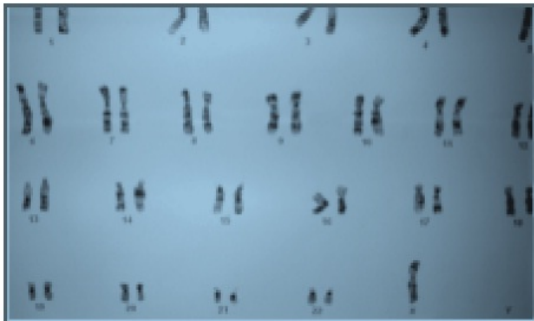


Fig. 4: Karyotyping : 45 XO cell line.

The combination of Turner's syndrome and an absent of uterus was very unusual. The patient & her family were informed of the unusual findings and she was started an oestrogen replacement therapy with incremental doses of ethinylestradiol. Because of the extremely unusual nature of her clinical findings a repeat ultrasonography scan performed following six months of oestrogen therapy. To our surprise the repeat ultrasonogram scan revealed a infantile uterus measuring about 37mm x 6mm. Further imaging after a period of oestrogen replacement is recommended if initial scans fail to identify any uterine tissue.

Discussion

Turner's syndrome is characterized by streak gonads within phenotypic females without somatic abnormalities. The existence of uterus is important in definition of gonadal dysgenesis. The absence of uterus has been regarded by a number of authors as a criterion for the diagnosis of androgen insensitivity syndrome.

There was absent of uterus in our patient with no secondary sexual characters. There was not a family history of primary amenorrhoea.

In our case gonadotropin levels was over the post menopausal level (FSH 36.50IU/L, LH 52.15 IU/L and estradiol was under the lowest measurable range (16.21pg/ml). The lack of menstruation opr breast development was related with insufficiency of gonadotropins.

Comment

The incidence of primary amenorrhoea is reported to be less than five percent of patient in a general gynecologic practice.⁹ it has also been reported¹⁰ that 43 percent of cases of primary amenorrhoea are cases of gonadal dysgenesis (turner's syndrome) or of testicular feminization.

After puberty, the diagnosis of gonadal dysgenesis is usally made when a female has primary amenorrhoea in conjunction with the absences of secondary sex characteristics and a variety of congenital defects.

It is generally agreed that women with gonadal dysgenesis should be encouraged to live normal lives and to indulge in normal marital relations. They should, however, se be advised that under the circumstances they will be infertile. The psychologic and physical well being of these patients can be helped greatly by the oral administration of oestrogens because of the ability of these hormones to develop and maintain the secondary sex characteristics. The patient in this case report has been put on this type of therapy with satisfactory results

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All correspondence to:
Munira Ferdusi
Assistant Professor
Department of Obs & Gynae
Rajshahi Medical College.