NORMAL MENSTRUATION IN A CASE WITH MANY OF THE MORPHOLOGICAL FEATURES OF TURNER'S SYNDROME – AN UNUSUAL MANIFESTATION

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SYNOPSIS

A case is reported of normal menstruation in a patient with many of the morphological features of Turner's syndrome.

SING MED J. 1989; No 30:105 – 106

INTRODUCTION

The Bonnivie-Ullrich-Turner (or Turner's) Syndrome, constituting about 3 in 10,000 full term births (1), is a problem of intersexuality with a chromosome pattern (karyotype) 45X0 determined at the time of conception. Since the Y chromosome is so powerful, in so far as the phenotype of the individual is concerned on account of its positive masculinizing force, its absence makes the Turner always appear female (2). However, the differential diagnosis of Turner mosaics can be an interesting intellectual exercise.

CASE REPORT

Mrs. R, a 38 year old woman married for four years was seen in the University Gynaecological clinic on 15 June 1984 complaining of a blood stained vaginal discharge of 3 months duration. This discharge, however, had been excessive for the past three weeks. The menarche was at 15 years with regular 30 day cycles the flow lasting 5 to 6 days. For the past four years the flow had been more excessive and the periods had also been painful. She had not been successful in achieving a pregnancy even though she had not used any contraception. Her height was 150 cm (5 feet) and her weight was 40 Kg. Other very striking features in this patient were the webbing of her neck, increased carrying angle (cubitus valgus), chest deformity (pectus cavum), widely spaced nipples and deformities of her toes. The cardiovascular and respiratory system were clinically normal. The blood pressure was 100/80 mm Hg. The development of secondary sexual characteristics, except for underdeveloped breasts, appeared to be reasonably adequate with satisfactory development of sexual hair (pubic and axillary). The external genitalia was typically female. On vaginal examination a small mass was felt protruding through the external os: the uterus was normal in size, anteverted and mobile. No adnexal or other masses were felt through the fornices. Speculum examination confirmed a polyp protruding through the external os. It was of moderate size and showed slight bleeding from its surface. The patient was admitted to the University Gynaecological Unit for further investigations and management. A buccal smear revealed that she was chromatin positive. Her visual acuity and colour vision were normal. The output of urinary 17 ketosteroids was slightly reduced (2.4 mg per 24 hours). A polypectomy and D & C was performed on 24.06.1984 and the patient discharged a few days later. The histology of the polyp and the curettings was reported as a myomatous polyp covered by endometrium and premenstrual endometrium respectively. Though requested to report at the Gynaecological clinic for further follow up she defaulted attendance.

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DISCUSSION

The striking clinical feature here is the finding of normal menstruation in a patient with many of the morphological/somatic features of the Turner's syndrome, but without the characteristics chromatin negative buccal smear.

"Turner's syndrome" means different things to different writers. First described by Turner (1938) the syndrome included short stature, sexual infantilism, cubitus valgus, shortening of the fourth metacarpal and webbing of the neck. Typical patients seldom attain a height of 5 feet. A broad shapeless trunk, deformities of the chest (pectus cavum), ears, wrists, fingers and toes, bone changes including osteoporosis, a low occipital hair line, exaggerated epicanthic folds and multiple naevi on the skin are the other physical abnormalities often, though not invariably, present. Congenital cardiac lesions, particularly coarctation of the aorta, congenital malformation of the kidneys, defects in visual acuity and colour blindness

have also been reported. Moreover streak gonads are also a feature of this syndrome. Most of these patients have a negative buccal smear (chromatin negative) and two thirds of them have a 45X0 karyotype. Others will be mosaics with a karyotype XO/XX, XO/XY and XO/XYY. In Turner mosaics with a karyotype XO/XX, between 5 and 15 percent of cells may be chromatin positive. Those with the karyotype XO/XY may show variable degrees of virilism and these individuals are better classified as cases of mixed gonadal dysgenesis (3). It is stated that about 21 sex chromosome complements have been associated with streak gonads, but only about 9 sex chromosome complements have been associated with Turner's syndrome (4).

The Gynaecologist would use the term Turner's syndrome to indicate sexual infantilism with ovarian streaks, short stature and two or more of the morphological abnormalities referred to above. Since some patients with the morphological characteristics of the Turner syndrome have been shown to have a normally positive X-chromatin body count. It is possible that our patient belonged to that category or, more likely, to that of a Turner mosaic. Although most mosaics are sexually infantile with a female phenotype some may menstruate (3). The possibility of a mosaic background explains why 20 percent women showing a fairly typical Turner's syndrome have chromatin positive buccal smears and Jeffcoate (2) observes that the situation is further complicated by the fact that 46XX/45XO mosaicism can, rarely, result in a functional ovary on one side and a streak gonad on the other resulting in a menstruating woman with the physical stigmata of Turner's syndrome.

Because the ovaries are inactive in the Turner the secretion of oestrogens is low but of FSH unusually high. The output of 17-keto steroids may be slightly reduced (2). Only the 24-hour urinary 17 keto steroids could be determined in our patient and this was found to be within the normal range, lending support to the possibility of our patient belonging to the category of an otherwise normal patient with apparently normal ovarian function, but possessing many of the morphological features of the Turner.

Although a future laparotomy was planned with a view to obtaining a biopsy from each gonad to ascertain its true nature this was not possible since our patient defaulted attendance after the initial surgery. Gonadal biopsy might have provided us more useful information particularly concerning the presence or absence of a streak gonad. It is worthy of emphasis that patients with XY mosaicism should have the area of gonadal streak removed to prevent the risk of development of a dysgenetic tumour (5). However, since our patient menstruated normally she could possibly not have possessed the karyotype XO/XY, even if she was a Turner mosaic but since gonadal biopsy was not performed the possibility of one functional ovary and one streak gonad in this patient cannot be altogether ruled out.

ACKNOWLEDGEMENTS

I thank the Director, General Hospital Jaffna (Teaching) for permission to publish this case and Miss. N. Sinnadurai of my Department for the typescript.

REFERENCES

1. Barnes J. Lecture Notes on Gynaecology. 4th ed. Oxford London Edinburgh Melbourne, Blackwell Scientific Publications 1980: 1-22.
2. Jeffcoate N. Principles of Gynaecology. 4th ed. London & Boston, Butterworths 1975: 155-86.
3. Shearman RP. In: Dewhurst J. ed. Integrated Obstetrics & Gynaecology for Postgraduates. London, Blackwell Scientific Publications 1981: 49-55.
4. Benson RC. Current Obstetric & Gynaecologic Diagnosis & Treatment. Lange Medical Publications 1984: 161-80.
5. Teter J. Boczkowski K. In: Dewhurst J. ed. Integrated Obstetrics & Gynaecology for Postgraduates. London, Blackwell Scientific Publications 1981: 49-55.