Successful Pregnancy in Turner Mosaic

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Turner syndrome or mosaic should be suspected in any prepubertal female with proportionate shortness of stature and primary amenorrhoea. We present an interesting case of successful pregnancy in a female with short stature and primary amenorrhoea confirmed to be Turner mosaic on karyotyping.

Case Report

A 15 year old girl was referred to us by a general practitioner in view of short stature and lack of development of secondary sexual characters. The development of milestones was normal and she had a good intelligent quotient (IQ). She was 136 cms tall, weighed 24 kgs with normal skin and hair line. There was slight webbing of the neck. There were no other Turner Stigmata. The breasts were Tanner Grade I and pubic hair showed Tanner Grade I distribution. Cardiovascular and respiratory systems were normal. External genitalia were infantile on examination.

Her investigations revealed low levels of estradiol 17.60 pg/ml (50-100 pg/ml), and low gonadotropins FSH 5.20 m IU/ml (0-20), LH – 0.20 MIU/ml (0-2) with normal T3, T4 and TSH. A buccal smear at this time was positive for X chromatin. A pelvic ultrasound showed a hypoplastic uterus with normal ovaries. The left ureter was dilated with hydronephrotic left kidney. Her kidney function tests were otherwise normal. Pyelography confirmed a non obstructive left hydroureteronephrosis. So a palliative uretero neocystostomy was done on the left side. A differential diagnosis of delayed puberty, or idiopathic gonadotropin deficiency was entertained until a karyotype was done. CTG banding revealed the diagnosis of Turner mosaic (few cell lines of XO). She was subsequently put on cyclical conjugated estrogen (Tab Premarin 1.25 mg OD x 21 days and Medroxy progestrone acetate (Tab Farlutal 5 mg x 1 week) on regular basis. Over 3 years there was improvement in her growth. Her height increased by 5 cm, the breast development showed Tanner Grade III and pubic and axillary hair growth showed Tanner Grade III. Within five years of cyclical hormones, she resumed menstruation, and later mensturated even without hormones. She was lost to follow up and presented 4 years later with 2 months amenorrhoea. Her urine pregnancy test was positive and ultrasound scan confirmed a viable pregnancy. She was booked at the high risk clinic with regular follow up. Pregnancy progressed uneventfully till 32 weeks, when she had recurrent attacks of urinary tract infection with fever. Serial ultrasound showed normal fetal growth with mild to moderate maternal hydronephrosis on left side. Antibiotics were given to treat UTI and her kidney function tests were maintained well. Elective lower segment caesarean section was done at 38 weeks for flexed breech presentation giving birth to a female child. The female baby weighed 2.8 kg with normal Apgar scores.

This case has been reported to highlight that buccal smear is not an acceptable method of screening for Turners, because at least 34% of cases are X chromatin positive. On the other hand buccal smear should be done along with chromosomal analysis on one or more tissues