A Rare Case of Primary Amenorrhea with Sex Chromosomal Mosaicism and Trisomy

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The incidence of trisomy 47XXX is approximately 1 to 1.5 per thousand live born females. These were originally described as super females, which they are not, as they are likely to suffer from oligomenorrhoea, secondary amenorrhea, infertility and genital tract hypoplasia. Again majority of women with a 47XXX karyotype appear to be normal in every way and are fertile. This taint is not transmitted to offpsring, the extra X being lost during oogenesis.

The etiology of trisomy and mosaic cell line is uncertain. If malsegragation or maldivision takes place at a later stage of development with retention of the initial cell line (normal 46XX) a mosaic cell line will be formed. This leads to presence of the abnormal cell line in the ovaries, thereby leading a to a diminished gonadal potential. This generally causes oligomenorrhea, secondary amenorrhea after a few initial normal cycles.

Interestingly the present case presented with primary amenorrhea with 46XX/47XXX mosaic cell line.

A young woman 23 years of age came with primary amenorrhea. She had normal height and weight. Her psychological and intellectual set up was normal.

Physical examination revealed a normal female phenotype with blood pressure : 130/84mm of Hg, anemia : nil, heart / lungs : normal, breast: Tanner III, height / span - 5'3"/5'6", P/abdomen : normal, P/R: Uterus anteverted normal size, Fx-clear.

Investigations revealed normal ultrasound pelvic structure. Hormonal assays revealed high levels of serum FSH and LH (S.FSH = 33.0 mlu/ml and serum LH = 21.6 mlu/ml), serum prolactin 20.5ng/ml, serum 17.0 Beta Oestradiol 12.0 pg/ml(normal 30.0 to 100), serum T_3, T_4, TSH were normal.

The patient responded to hormonal withdrawal therapy. (Oestrogen/progestogen combination).

Cytogenetic analysis was carried out based on banding technology done on 100 well spread metaphase chromosome fields. No change in the form of structural aberration had been detected. But, change in the form of numerical aberration had been detected. i.e., 80% of the cell line was 47XXX and 20% of the cell line was 46XX. Sex chromatin study on buccal mucosa showed double chromatin approximately 80% of 30% Drumstick in polymorphonucleus shows single in 3% cases.

Due to abnormality of sex chromosomal complement, the ovaries failed to respond to FSH and LH secreted by the pituitary. Absence of negative feedback by estrogens from target tissue led to high level of serum FSH and LH. All these figures clinical, hormonal and cytogenetic analysis together were suggestive of primary ovarian failure due to chromosomally incompetent structure.

She was put on hormone replacement therapy with progesterone and low dose oestrogen.

Hormonal preparation of endometrium with ovum transfer (donation) remains the only remote possibility of her future fertility.