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Research Letter

Secondary amenorrhea associated with 46,X,der(X)t(X;X)(p22;p22)

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Translocation involving an X chromosome is rare, moreover, translocation between two X chromosomes are rarer occurrences. They exist as 46,X,t(X; X) or 45,X/46, X, dic(X). Most frequently, a duplication of almost the entire X chromosome occurs with a deletion of a part of the arm at the breakpoint. Such duplication/deficiency of X chromosome material leads to abnormal positioning of genes, monosomy for one part of X and trisomy for another. The phenotypes range from Turner syndrome to only ovarian dysgenesis without other Turner syndrome stigmata [1,2].

The proband was a 20-year-old phenotypic woman. She was consulted in the Genetic center because of short stature and secondary amenorrhea. The patient was from a full-term delivery and was born to a 31-year-old father and a 26-year-old mother with a consanguineous marriage. (The pedigree is shown in Fig. 1). There was no history of fetal wastage and patients' two brothers are healthy.

The proband had an uncomplicated antenatal, perinatal, and neonatal course. Her birth weight was 1,500 g and there was no lymphedema noted during early infancy. She had been small since early childhood, and her psychomotor development was normal. She first noted some breast development at age of 14. One year later, she experienced her first menstrual period, which lasted for 3 days. However, she has had no further menstruation.

The physical examination at 20 years of age showed a short and rather obese, but quite intelligent. Her height was 143 cm and her weight 95 kg. Furthermore, she had several stigmata suggestive of Turner's syndrome, i.e. triangular facies with small mandible, low hairline, broad shoulders, widely spaced nipples and short forth metacarpals. Her breast development was minimal. She had sparse, curly pubic and axillary's hairs. Her blood pressure was normal.

Abdominal ultrasonography of patient showed bilateral ovarian hypoplasia.

Biochemical tests such as FSH and LH showed increase in their level .The thyroid function test and the level of prolactin were normal.

Her karyotype was analyzed using a conventional banding technique (GTG). Analysis of 25 metaphase cells showed 46,X,der(X)t (Xqter Xp22::Xp22 Xqter) in all cells (Fig. 2) [3]. Her mother was found to have a normal karyotype.

The formation of this X/X translocation could be the result of breakage and rearrangement between two X chromosomes or of a crossover in a pericentric inversion loop during the first meiotic division in a preceding generation [4].

In 1961, Edwards [5] reported the first case of possible X/X translocation in a phenotypically normal female with primary amenorrhea.

Tarkan et al [6] reported a 46,X,t(X;X)(p22;p22) karyotype for the first time in the chromosome analysis of a 22-years-old woman patient with secondary amenorrhea.

Our case is the second reported instance of a patient with ovarian hypoplasia that involves translocation between two X chromosomes in region of p22;p22.

Kim et al [7] studied a family with an X/X translocation in region of p22;q13 which was found in a 16-year-old daughter and in her 34-year-old mother. The daughter had short stature,

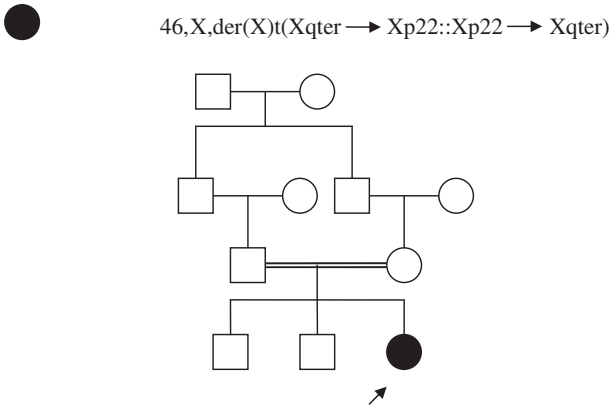

Fig. 1. Pedigree of the family.

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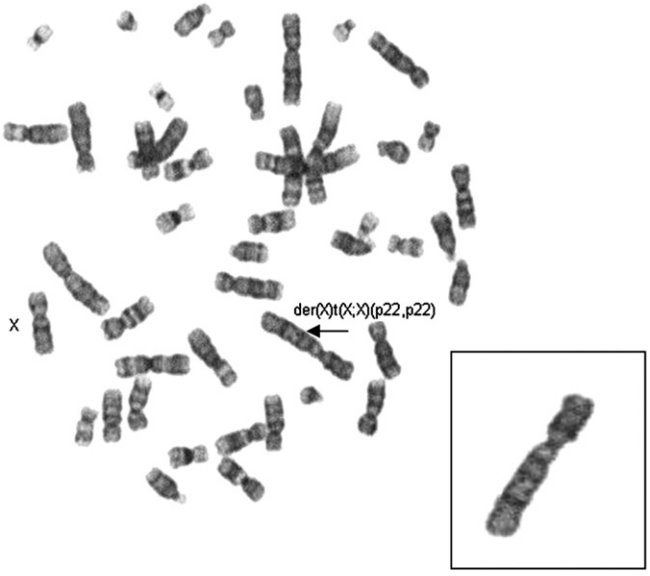

Fig. 2. karyotype of the proband.

secondary amenorrhea and many Turner's stigmata. Furthermore, the mother had a few Turner's stigmata and had developed secondary amenorrhea at the age of 24. In our case, the patient had secondary amenorrhea, turner's stigmata with a 46,X,der(X)t (Xqter → Xp22::Xp22 → Xqter) karyotype; whereas her mother had no turner's stigmata with a normal karyotype.

Gerard [8] reported a patient with delayed puberty and a previously unreported translocation 46,X,der(X)t(X;X)(q22; p11.2) without any manifestations of turner syndrome.

Rivera et al [9] explained a girl with short stature, primary amenorrhea, mild turner stigmata with a 45,X/46,X,t(X;X) (p22.3;p22.3) karyotype.

Ferdinand Sauer et al secondary amenorrhea associated with translocation of a part of the long arm of an X chromosome to the short arm of a number 7 autosome in a girl with gonadal dysgenesis reported. They stated the importance of considering gonadal dysgenesis early in the evaluation of patients with secondary amenorrhea considered for ovulation induction is stressed [10].

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