Case Report

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Monozygotic Twinning in a Female with Triple X [47, XXX]

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Key Words

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Abstract

We report a successful twin pregnancy in a woman with secondary amenorrhea who is genotypically 47, XXX. Cytogenetic markers suggested that they are apparently monozygotic twins.

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In 1959, Jacobs et al. [1] first described a female with 47, XXX, an anomaly which is now recognized to be as frequent as 1/1,000. Nevertheless, it is not considered a clinical syndrome in the 'true sense'. Fertility is normal although menstrual disorders are frequently seen [2-5]. To the best of our knowledge, a twin pregnancy in this disorder has not been reported.

A 37-year-old Caucasian female was referred for chromosomal evaluation due to secondary amenorrhea. The patient's menarche occurred at the age of 14 years. Chromosome preparations were obtained from peripheral blood using standard methods [6]. Cytogenetic findings with G-banding revealed an abnormal 47, XXX karyotype in 100 cells analyzed. One year later, when she was 20 weeks pregnant, she returned to our laboratory for amnio-centesis. A twin pregnancy was observed by ultrasound. While pregnant, she took three courses of Provera and Premarin for withdrawal of bleeding. Other clinical and laboratory findings are unremarkable.

Since the proband was at high risk to produce progeny with aneuploidy for X-chromosome, the uncultured am-niocytes were stained immediately by the FISH technique [7] using the X-chromosome-specific probe for rapid diagnosis (DXZ1, Oncor, Gaithersburg, Md., USA). The presence of two centromeric dots at interphase nuclei from each twin was an apparent indication of normal twins with 46, XX karyotypes (fig. 1). Chromosome anal-

Fig. 1. Interphase nuclei stained by FISH technique showing three dots in the patient (A) while in twins (B, C) two dots were seen. A metaphase from the patient is shown in D where all three X-chromosomes are marked (see text).

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Fig. 2. The most distinct heteromorphous markers on chromosomes 3 and 7 revealed by y4M/Giemsa technique are shared by both twins suggesting an apparent monozygosity (see text). ysis was performed from amniocytes of both sacs and confirmed the initial impression. A series of selective staining techniques performed on metaphases revealed identical cytogenetic markers shared by the twins, suggesting they were identical (fig. 2). Obviously, a single normal egg was fertilized, followed by zygote splitting. There is one report where a twin pregnancy was noted in

a woman with mosaic karyotype of 45, X/47, XXX [8]. However, this is the first report of a twin pregnancy in a woman with secondary amenorrhea and a 47, XXX karyotype.

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References

- Jacobs PA, Baikie AG, Court-Brown WN, MacGregor TN, Maclean N, Harnden DG: Evidence for the existence of the human super-female. *Lancet* 1959;ii:423-425. Telfer MA, Richardson CE, Helmken J, Smith GF: Divergent phenotypes among 48, XXXX and 47, XXX females. *Am J Hum Genet* 1970; 22:326-335.
- Dewhurst J: Fertility in 47, XXX and 45, X patients. *J Med Genet* 1978;15:132-135.
- de Grouchy J, Turleau C: *Clinical Atlas of Human Chromosomes*. New York, Wiley, 1984, pp 1-384.
- Ballesta F, Zapata M: Phenotype in triple-X patients. *Clin Genet* 1989;34:388-389. Verma RS, Babu A: *Human Chromosomes: Manual of Basic Techniques*. New York, Pergamon Press, 1989.
- Pinkel D, Landegent J, Collins C, Fuscoe J, Segraves R, Lucas J, Gray J: Fluorescence in situ hybridization with human chromosome-specific libraries. *Proc Natl Acad Sci USA* 1988;85:9138-9142.
- Kivinen S, Herva R: Twin pregnancy in a woman with 45, X/47, XXX karyotype. *Obstet Gynecol* 1980;56:401-402.

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Twinning in Triple X