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Chimerism in a fertile woman with 46,XY karyotype and female phenotype

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Abstract

We report on the unexpected finding of a 46,XY karyotype in a 30 year-old woman with normal ovarian function and a former pregnancy at 17 years of age. Chromosome analysis was performed prior to intracyoplasmic sperm injection (ICSI), due to infertility of her husband. Repeated chromosome analysis in lymphocytes of the female resulted in a normal male karyotype. Fluorescence in-situ hybridization (FISH) analysis of cultured lymphocyte interphase nuclei detected in 99% of the cells one X and one Y chromosome-specific signal respectively, whereas two X chromosome-specific signals were observed in only 1% of the nuclei. Chromosome analysis of fibroblasts of ovarian and muscular tissues as well as of skin revealed a normal female karyotype (46,XX). Chimerism could be proven by variable number of tandem repeats (VNTR) analysis. Since the case history of the patient revealed that her twin brother died shortly after birth, it can be assumed that chimerism is caused by feto-fetal transfusion during pregnancy and delivery of the proposita.