Taiwanese Journal of Obstetrics & Gynecology 56 (2017) 410–411

Contents lists available at ScienceDirect

Taiwanese Journal of Obstetrics & Gynecology

journal homepage: www.tjog-online.com

Research Letter

Molecular cytogenetic characterization of mosaicism for a small supernumerary marker chromosome derived from chromosome 7 in the male partner of a phenotypically normal couple with repeated spontaneous abortions

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ARTICLE INFO

Article history:
Accepted 30 March 2017

Dear Editor,

A non-consanguineous infertile couple was referred for genetic counseling because of two spontaneous abortions. The couple had two natural conceptions ending in spontaneous abortions. The wife was 36 years old, and the husband was 41 years old. The woman did not have any child and had undergone assisted reproductive technology because of infertility. The urological, gynecological and infectious investigations of the couple revealed normal results. Cytogenetic analysis of the couple' peripheral bloods revealed a karyotype of 46,XX in the wife and a karyotype of 47,XY+mar[14]/46,XY[26] in the husband. Among 40 cultured lymphocytes derived from the husband's peripheral blood, 14 cells had a karyotype of 47,XY,+mar (Fig. 1), while the other 26 cells had a karyotype of 46,XY. Metaphase fluorescence *in situ* hybridization (FISH) analysis on the small supernumerary marker chromosome (sSMC) using the chromosome 7 α-satellite specific gene probe (D7Z1, 7p11.1-q11.1) [spectrum green, fluorescein isothiocyanate (FITC)] and the chromosome 6 α-satellite specific gene probe (D6Z1, 6p11.1-q11.1) (spectrum red, Texas Red) (Cytocell Inc. Adderbury, Oxfordshire, UK) showed that the sSMC was derived from chromosome 7 and the result of 47,XY,+mar.ish der(7)(D7Z1+) in the sSMC (Fig. 2).

The sSMC has been implicated in human infertility [1–3]. In a study of 470 infertile couples referred for assisted reproductive technique, Marchina et al. [1] found chromosome aberrations in 2.55% of the couples. According to the report of Marchina et al. [1], the female partners had low-level sex chromosome mosaicism in 1.28% and structural autosomal abnormalities in 1.06% of the cases, and the male partners had autosomal abnormalities in 1.49%, sSMCs in 0.42% and Y chromosome inversions in 0.42% of the cases. Manvelyan et al. [2] found an sSMC rate of 0.125% in patients with fertility problems and an sSMC rate of 7% in males with decreased sperm parameters. In a study of 234 infertile patients as carriers of sSMCs, Armanet et al. [3] found that 72% had the sSMC originated from acrocentric chromosomes, 30% had euchromatic imbalance, and 1.2% had identified putative genes. Olszewska et al. [4] suggested a genetic dosage and position effect of sSMC in human nuclei in infertile male patients. Armanet et al. [3] suggested a gene

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http://dx.doi.org/10.1016/j.tjog.2017.04.029

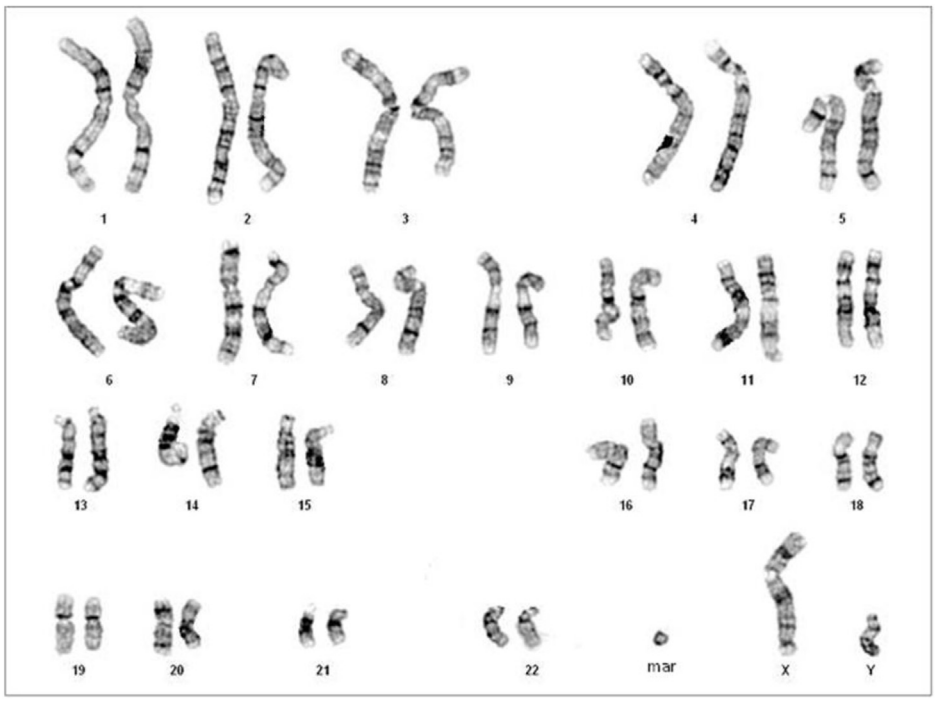

Fig. 1. A karyotype of 47,XY,+mar. mar = marker chromosome.

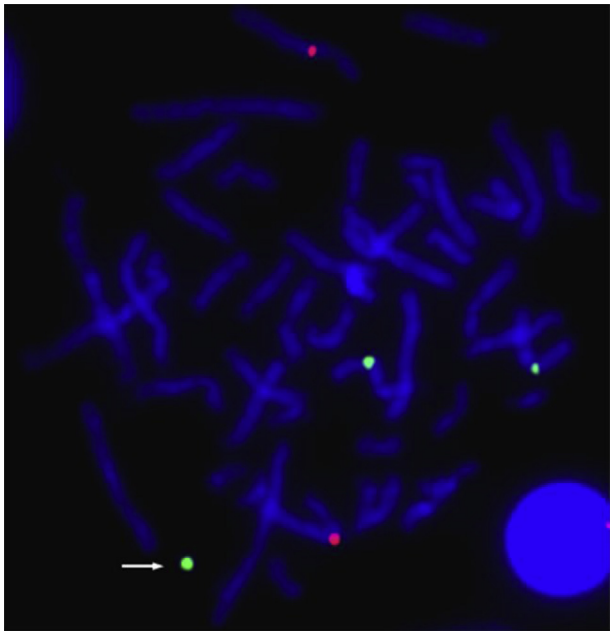

Fig. 2. Metaphase fluorescence *in situ* hybridization of the lymphocytes obtained from the husband's peripheral blood shows three green signals of chromosome 7 α-satellite specific probe (D7Z1, 7p11.1-q11.1) and two red signals of chromosome 6 α-satellite specific probe (D6Z1, 6p11.1-q11.1), indicating that the small supernumerary marker chromosome is derived from chromosome 7. The arrow indicates the marker chromosome.

dosage increase effect and mechanical effects perturbing meiosis of sSMC on human infertility. Balkan et al. [5] previously reported an sSMC derived from chromosome 22 in the male partner of a couple with repeated spontaneous abortions. We additionally report an sSMC derived from chromosome 7 in the male partner of a couple with repeated spontaneous abortions. Our case adds to the list of an sSMC in male partners in infertile couples associated with repeated spontaneous abortions.

Conflict of interest

The authors have no conflicts of interest relevant to this article.

Acknowledgements

This work was supported by research grants MOST-104-2314-B-195-009 and MOST-105-2314-B-195-012 from the Ministry of Science and Technology and MMH-E-105-04 from MacKay Memorial Hospital, Taipei, Taiwan.

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