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Case Report

A small supernumerary marker chromosome, derived from chromosome 22, possibly associated with repeated spontaneous abortions

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ABSTRACT. We report a phenotypically normal couple with repeated spontaneous abortions and without other clinical features. Clinical, hematological, biochemical, and endocrinological aspects of the couple did not reveal any abnormalities. The karyotype of the wife was normal (46,XX), while the husband was found to have an abnormal karyotype, 47,XY,+der(22)mat. The marker chromosome was familial and non-satellite. Although the potential risk of small supernumerary marker chromosomes for spontaneous abortions cannot be defined precisely, marker chromosomes, together with methods used for ascertainment, are also factors to be considered when investigating infertility consequences. Furthermore, identification of the origin of a marker chromosome may provide additional information for patient karyotype-phenotype correlations. Further studies, such as molecular analyses to identify the breakpoint, are necessary for investigating

phenotype-genotype correlations and assessment of genetic risks for small secondary chromosomes. The cause of repeated spontaneous abortions in this couple might be the presence of this marker chromosome in the husband. Consequently, we recommended genetic counseling before further pregnancies.

Key words: Small supernumerary marker chromosomes; Karyotype; Spontaneous abortion; Marker chromosome

