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47,XX + mar karyotype containing genes from the azoospermia factor region. A case report

Franco Causio [1], Ernesto Gentile, Rita Fischetto, Nicoletta Archidiacono, Nicola Magro

Affiliations + expand

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Abstract

Background: Abnormal embryo development is the major cause of implantation failure and accounts for the low rate of human fertility in vitro and in vivo. Chromosome abnormalities are widely involved in this process through meiotic nondisjunction, fertilization abnormalities and mitotic nondisjunction.

Case: In our assisted reproductive technology program a couple underwent cytogenetic analysis. The woman had a 47,XX + mar karyotype. We investigated this patient by chromosome analysis, fluorescence in situ hybridization (FISH) and polymerase chain reaction (PCR) DNA analysis. The marker chromosome was found to be very similar to a Y chromosome in size and QFQ staining pattern. Therefore, it was tested by FISH using alpha- and beta-satellite DNA specific for the Y chromosome, some YACs specific for the long arm of the Y chromosome and alpha-satellite DNA specific for 15 chromosomes as probes. In order to define this marker, the next step was PCR amplification of the whole genomic DNA using specific landmarks (sequence-tagged sites) to encompass the azoospermia factor (AZF) region on the long arm of the Y chromosome.

Conclusion: A woman had an extra chromosome containing centromeric DNA derived from the Y and 15 other chromosomes, heterochromatic regions derived from 15 chromosomes and a large heterochromatic block at the end of the long arm that definitely was not Y chromosome heterochromatin (beta-satellite). PCR showed several genes of the Y chromosome long arm that are assumed to be involved in male gametogenesis. Phenotypic effects could not be excluded because of the presence of AZF genes. Oocyte karyotyping might better explain the role of the genetic problem on female infertility.