Online journal ISSN - 1676-5680
GMR Genetics and Molecular Research
EVOLUTION AND TECHNOLOGY

Tracking microdeletions of the AZF region in a patrilineal line of infertile men

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Genet. Mol. Res. 7 (3): 614-622 (2008)
Received May 6, 2008
Accepted June 18, 2008
Published July 8, 2008

ABSTRACT. Male infertility is considered to be a difficult-to-treat condition because it is not a single entity, but rather reflects a variety of different pathologic conditions, thus making it difficult to use a single treatment strategy. Structural alterations in the Y chromosome have been the principal factor responsible for male infertility. We examined 26 family members of 13 patients with male infertility who showed deletions in the AZF region. In family 1, the father and a brother did not show microdeletions. However, a son showed a microdeletion in AZFa (sY84) and an azoospermic sperm analysis, but another son had a microdeletion in AZFa (sY84) and AZFb (sY127) and a normal sperm analysis. The father of family 2, with severe oligozoospermia, had a microdeletion in the AZFa region (sY84) and his son, conceived by intracytoplasmic sperm injection, also showed the same microdeletion. In the other families, only the men with an altered sperm analysis had a microdeletion. It is possible that in family 1, the father and brother who did not show microdeletions in this study, could have microdeletions in regions upstream or downstream of the one analyzed. The treatment with intracytoplasmic sperm injection can result in vertical transmission of microdeletions of the AZF region and can also cause the expansion of a *de novo* mutation. This finding reinforces the necessity of an

investigation of microdeletions of the Y chromosome in individuals who are candidates for assisted reproduction, as well as genetic counciling and follow-up.

Key words: Male infertility; Y chromosome; Vertical transmission

