Comments on the AZFc markers used for screening of Yq microdeletions

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To the Editor:

We have read with interest the article by Balkan and colleagues [1] in your November issue that screened Y chromosome microdeletions in infertile males with oligozoospermia and azoospermia in Southeast Turkey. We would like to comment on one false positive result and discuss some of the markers that they used for screening of microdeletions in AZFc region. We believe that it could be useful for the Journal of Assisted Reproduction and Genetics readers.

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First, they described one azoospermic patient (Md-58) that was negative for the two markers sY277 and sY153, while positive for sY152. They stated that sY277 is inside of the *DAZ3* gene. We have defined the relative location of sY277, sY254, sY255, sY153 and sY152 using primer blast program available at the University of California, Santa Cruz (UCSC) Genome Browser on the MSY (male-specific region of the human Y chromosome) reference sequence [5]. The marker sY277 is located within the *DAZ* genes and there is at least one copy inside of each *DAZ* gene. Negative results for the sY277 indicate a large deletion which includes all copies of the *DAZ* genes (Fig. 1). Consequently, given the relative position of these markers as clearly depicted by Fig. 1, deletion of sY277 without deletion of sY152 is impossible and should be regarded as a methodological error.

Second, they surprisingly stated that the two markers sY254 and sY255 are inside of the *DAZ1* and *DAZ2* genes, respectively. Four *DAZ* genes are 99.9% identical and there is at least one copy of each marker inside of each *DAZ* gene (Fig. 1) [3].

Third, the three markers sY145, sY152 and sY153 which they referred to AZFd, are actually inside of AZFc region and there are at least two copies of each marker in this region. Nowadays, it is accepted that putative AZFd region does not exist [2, 4].

Finally, to avoid these technical flaws there is validated guideline which could detect up to 95% of all AZF microdeletions [4].

Fig. 1 Schematic diagram shows relative position of the markers sY152, sY153, sY254, sY255 and sY277. The figure clearly shows that sY152 is located in the deleted region. The highlighted markers are

not recommended by the European Academy of Andrology (EAA) and the European Molecular Genetics Quality Network (EMQN)

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