

Correspondence

Methodological errors in screening of Yq microdeletion in Iranian azoospermic men

Sir,

There are some discrepancies in the prevalence of Y chromosome microdeletions among Iranian azoospermic men reported by various researchers. We read with interest the article by Mirfakhraie *et al*¹ and noticed some methodological errors which cast doubts on the validity of their results.

They reported that the prevalence of Yq microdeletions among Iranian azoospermic men was 12 per cent and azoospermia factor region AZFb had the most common microdeletion (66.67% of total deletion) followed by AZFc (41.67%)¹. This is perhaps the first study reporting that the prevalence of AZFb microdeletion was higher than AZFc. Based on a meta-analysis of data on the Y chromosome microdeletions, relative prevalence of AZFc deletions is reported to be the highest (59.6% of total deletion) followed by AZFb microdeletions (15.8)². Mirfakhraie *et al*¹ described three patients who were negative for sequence tagged site (STS) makers sY254, sY255, sY239 and sY242. On the other hand, analysis of AZFd in these patients showed absence of sY145 and presence of sY153¹.

The absence of sY254 and sY255 indicates complete deletion of AZFc region³.

To determine the relative location of the mentioned STS markers, we used *in silico* PCR which is available at the University of California Santa Cruz (UCSC) Genome Browser. The sY145 and sY153 are present in three and two copies, respectively (Fig.). Regarding the position of these STS markers on the MSY (male-specific region of the Y chromosome) reference sequence⁴ (Fig.), presence of sY153 STS marker in the cases with deletions of the AZFc region and sY145 is not possible and should be considered as a methodological error.

Mirfakhraie *et al*¹ reported one patient with complete deletion of AZFc region, while the sY153 and sY145 were present. Theoretically, according to the relative positions of these STSs (Fig.), the deletion of sY145 and sY153 in patients with deletion of AZFc is possible. However, Noordam *et al*⁵ showed that all AZFc deletions would be negative for all AZFd STSs (except for sY152 which is inside DAZ genes).

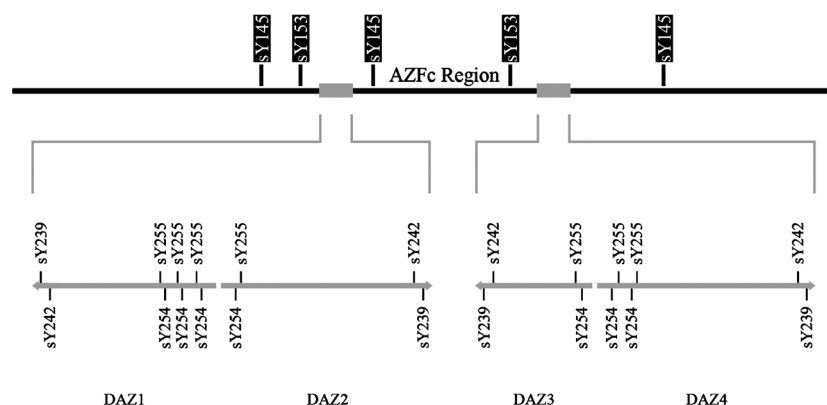


Fig. Relative position of STSs inside AZFc and putative AZFd regions used by Mirfakhraie *et al*¹. The STSs of the putative AZFd region are highlighted in black.

Besides these methodological errors, the sequence of MSY and the mechanism of Yq microdeletions have definitely shown that putative AZFd does not exist^{6,7} and STS markers referred to as AZFd are actually inside AZFc region³.

Finally, for detection of AZF microdeletions there are validated guidelines endorsed by the European Academy of Andrology (EAA) and the European Molecular Genetics Quality Network (EMQN) which could detect up to 95 per cent of all reported AZF microdeletions⁷.

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