Authors response

Sir,

We appreciate the authors¹ for their comments on our AZFb microdeletion in Iranian patients with idiopathic non-obstructive azoospermia².

As mentioned in their comments, the European Academy of Andrology (EAA) and the European Molecular Genetics Quality Network (EMQN) endorsed useful guidelines for the better detection of AZF microdeletions³. Our study was designed according to the EAA and EMQN guidelines, and we used the same STS markers suggested by these guidelines. Other STS markers were selected according to the previous reports and also the reviewers' comments. The absence of unamplified STS markers was confirmed by two additional PCRs.

Although deletions in the AZFc region are the most commonly reported deletions among AZF regions, but it should not be considered as an uncompromising rule. For example, at least there is one report on Y chromosome microdeletion in Turkish infertile men with the same frequencies for deletions in AZFb and AZFc regions⁴.

According to the authors' comments, using protocols suggested by EAA and EMQN one could detect up to 95 per cent of all reported AZF microdeletions^{1,3}. However, it seems that the validity of the detection frequency may vary in different populations. For example, using only sY84 and sY86 STS markers, Thangaraj *et al*⁵ have diagnosed patients who possessed deletions in the AZFa rejoin as normal individuals. They found deletion of sY746 in six patients and concluded that some deletions may be more predominant in certain populations⁵.

It has also been mentioned that the putative AZFd region does not exist definitely. However, Kent-First *et al*⁶ suggested the presence of the AZFd region between AZFb and AZFc regions. The Promega kit, Y Chromosome Deletion Detection System, also includes the detection of the AZFd region (Promega, Madison, WI). In addition, deletions in this region have been reported as common among infertile men in several studies⁴⁻⁸. Müslümanoğlu *et al*⁸ considered AZFd as a separate region in their study, described samples that were negative for sY254 and sY255 markers, while positive for either of sY145 or sY153 STSs. This study along with several others also conflicts with this

idea that the absence of sY254 and sY255 indicates complete deletion of AZFc region^{4-6,8}.

It seems that at least sY153 is polymorphic and exists in multiple copies⁹. Referring to UniSTS database of sequence tagged sites in the National Center for Biotechnology Information (NCBI), sY153 marker matches more than two locations on the human genome. Simoni *et al*¹⁰ have considered this marker among STS loci that should not be used in the study of Y chromosome microdeletions because it is polymorphic. This fact may explain the observed discrepancies concerning this marker in our study.

Finally, we would like to emphasize once more that our study² only reports the findings of our research and the mechanisms involved in different types of deletions should be further studied.

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