Current status of the molecular diagnosis of Y chromosome microdeletions in the work-up of male infertility

Factors influencing the variable incidence of Y chromosome microdeletions in infertile patients

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The article by Simoni et al. (1998) which summarized the published studies on the presence of Y chromosome microdeletions in infertile male patients was very interesting. By reviewing this literature, the authors attempted to answer the following three fundamental questions on this continuously evolving issue: (i) what is the real frequency on microdeletions in infertile patients?; (ii) which microdeletions are clinically relevant?; and (iii) in which patients are molecular diagnosis indicated? We think that all can easily agree with the authors that only patients with a sperm concentration $<5\times 10^6$/ml should be screened for the presence of Y chromosome microdeletions, regardless of the concomitant presence of varicocele (as also suggested by our own personal experience), and that only discrete microdeletions never found in normal controls should be assigned a causative role in the pathogenesis of oligoazoospermia (Pryor and Roberts, 1998). However, it is much more difficult to have a common view on the incidence of patients exhibiting microdeletions of the Y chromosome. As pointed out by Simoni et al. (1998), this percentage varies among studies ranging from 1% (van der Ven et al., 1997) to 37.5% (Foresta et al., 1997). Albeit a different frequency of microdeletions in the various populations cannot be excluded, it is our opinion that at least two other factors may account for this variability. First there are no standardized selection criteria of the sample group. For example, the percentage of microdeletions in azoospermic subjects who should have been excluded since Y chromosome microdeletions are responsible for a quantitative, but not qualitative, spermatogenetic decline. Recalculating the data with this adjustment, the percentage of microdeletions rises to 13.3%, while the percentage of microdeletions in patients with idiopathic infertility reaches 17% instead of the reported 9.8%. We therefore suggest a more critical evaluation of the data published and propose to avoid the use of the term infertile men, which may be misleading, and to report the sperm concentration output and a complete andrological evaluation of the cases enrolled. Certainly, normozoospermic infertile patients should not be screened for azoospermia factor nor included in the sample group.

References


