Recombination in heterozygote inversion carriers

Sir,

We have read with interest the recent publication in Human Reproduction of Malan et al. (2006), a case report dealing with the evaluation of unbalanced sperm produced by a carrier of an inv(21)(p12q22.3).

In their work, the authors report a high amount of recombinant gametes (32.6%) from an inversion, which involves a segment of about 47 Mbp of chromosome 21 and represents around 90% of the total length of the chromosome. In their discussion, they refer to a model of behaviour for inversions proposed by our group (Anton et al., 2005) which considers the implication of the size and the proportion of the chromosome inverted in the production of recombinant gametes. Among their conclusions, they state that our model ‘could be true for long chromosomes but not necessary for the smaller ones’. We would like to point out that this conclusion was achieved by taking into account only a part of the model described by us, which only considers the length of the inverted segment as the single parameter to influence the production of recombinant gametes and does not take into account the effect of the proportion of such a segment within the affected chromosome.

To build our model, statistical correlation analyses were performed (Pearson correlation test, \(P < 0.0001\)) and a statistically significant correlation was found between the inverted segment size and the production of recombinant gametes \((R^2 = 0.8555)\) and also between the proportion of the chromosome implicated in the inversion and the presence of unbalanced gametes \((R^2 = 0.7397)\). Consequently, we stated that ‘the size of the inverted segments and their proportion in the chromosome are two parameters closely related with the incidence of recombination’ (Anton et al., 2005).

Additionally, it is well known (Kaiser, 1984) that in inversions involving most of the chromosome length (independently of the absolute length of the chromosome), synopsis would tend to prevail between the inverted segment and its homologous non-inverted segment over the terminal segments (Figure 1; Anton et al., 2005). Thus, in these cases, although no inversion loop would be formed, recombination within the inverted segment would be allowed as well, and the production of recombinant gametes would also be expected.

In the work of Malan et al. (2006), although the inversion is shorter than 100 Mbp (total length of chromosome 21 = 50 Mbp), it involves around 90% of the total chromosome length. Therefore, according to these characteristics, the results obtained in their segregation study would fit entirely within the model proposed.

We are aware that our review and thus the model were conditioned by the scarce presence of information in the literature regarding the segregation behaviour of inversions in small chromosomes. To our knowledge, only one segregation study had been reported (Jenderny et al., 1992) and no recombinant products were found. Malan’s study represents a valuable intake of data that will contribute to elucidate the importance of the proportion of the inverted segment on the production of unbalanced gametes. More studies in inversion carriers of similar characteristics would be of interest to clarify the role of the proportion and the absolute length of the inverted segment on the final outcome of recombinant gametes.

References


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The human sex ratio in New York City did not change after 11 September 2001

Sir,

In a recent article in this journal, Catalano et al. (2006) state in their summary that the sex ratio (odds of a male birth)