Reviewer’s report

Title: The R337H Mutation in TP53 and Breast Cancer in Brazil

Version: 1 Date: 22 February 2012

Reviewer: Cezary Cybulski

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Gomes et al evaluated the frequency of the R337H mutation in p53 in 390 breast cancer patients and 324 controls from Rio de Janeiro. Two breast cancer cases (0.5%) and none control carried the mutation. Both cases had an early age at diagnosis (<40 years old) and a family history of breast and other cancers. This is the first study of R337H founder mutation in unselected cases with breast cancer in Brasil and certainly deserves publication!

Comments

Results:
Please draw family trees for the two mutation positive families.

Discussion:
The authors conclude that the frequency of R337H founder mutation (0.5%) in their study is lower then in two previous studies in other regions of Brasil (Sao Paolo and Porto Alegre) which reported frequency of 2.4% (San Paolo) and 11% (Porto Alegre), and they suggest that there may be regional differences in the mutation frequency as Sao Paolo and Porto Alegre are 442 km and 1546 km from Rio de Janeiro. This conclusion is wrong as it is derived from comparison of different series of cases - in Rio de Janeiro unselected cases of breast cancer were studied, in Sao Paolo a mix of high risk families with multiple breast cancer cases (n=45) and sporadic cases of breast cancer (n=78) was analyzed, and in Porto Alegre, Li-Fraumeni and LFS-like families (only some presented with breast cancer) were genotyped. It is not surprising that the frequencies in these series are different. You can not compare apples to pineapples because they sound similar.

Level of interest: An article of importance in its field

Quality of written English: Acceptable

Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:

no conflict of interests