http://dx.doi.org/10.3346/jkms.2012.27.3.336 • J Korean Med Sci 2012; 27: 336

The Author Response

Presence of Evolutionary Pressures or Genotyping Error

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I would like to thank the interest and comments to our paper entitled "XRCC1 polymorphisms and risk of papillary thyroid carcinoma in a Korean sample" (1). In our study, we genotyped XRCC1 Arg194Trp and Arg399Gln single nucleotide polymorphisms (SNPs) in 111 papillary thyroid carcinoma patients and 100 normal control subjects. In the results of our study, the XRCC1 Arg194Trp Arg/Trp genotype was significantly associated with a decreased risk of papillary thyroid carcinoma compared to that of Arg/Arg genotype, while the XRCC1 Arg399Gln genotype was not associated with a risk of papillary thyroid carcinoma.

I agree with the comments. There was a mistake in the calculation of Hardy-Weinberg equilibrium (HWE). A genotype distribution of XRCC1 Arg194Trp was in HWE (control P = 0.728, case P = 0.768); however, a genotype distribution of *XRCC1* Arg399Gln deviated from HWE (control P = 0.0002, case P =0.0003). The deviation from HWE could be associated with the sampling bias or the presence of evolutionary pressure, and it might be also associated with the genotyping error. As I already mentioned some limitations of our study in the paper, there was a selection bias as a hospital-based case-control study. The control group in our study may not have accurately represented the general population, and the genotype distribution of XRCC1 Arg399Gln deviated from HWE. Also, our study included a relatively small number of cases and controls. Therefore, the results of our study should be interpreted with caution. A further study with using large, matched case-control samples is necessary to validate the genetic effects of XRCC1 polymorphisms in the Korean population.

REFERENCE

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