CHEK2 c.444+1G>A variant and papillary thyroid cancer – own study and meta-analysis

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Background. Genetic predisposition to papillary thyroid cancer (PTC) is known to be muligenetic and complex with many genes interacting with environmental factor. However, the genes responsible for this predisposition mostly are not known. Many of them were analysed, but only for some genes the association with thyroid cancer is well established. Among analysed genes was CHEK2, but with the relatively small number of 468 PTC cases included. The aim of our study was to analyse the association of c.444+1G>A (formely IVS2+1G>A) CHEK2 variant in the big number of 2279 of PTC cases and 1218 controls. c.444+1G>A variant was analysed with HRM methods and confirmed by Sanger sequencing. The second purpose of the study was to perform the meta-analysis in all available Polish data to summarize c.444+1G>A CHEK2 variant association with PTC.

Results. A significant association was seen for c.444+1G>A with OR=4.49. Performed meta-analysis have confirmed these results - for c.444+1G>A association was seen with OR=5.89.

Conclusions. We have confirmed the association of c.444+1G>A CHEK2 variants with PTC in Polish population.

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