Prevalence of the E318K and V320I *MITF* germline mutations in Polish cancer patients and multi-organ cancer risk--a population-based study

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The E318K mutation in the MITF gene has been associated with a high risk of melanoma, renal cell carcinoma and pancreatic cancer, the risk of other cancers have not been evaluated so far. Herein we examined possible association of E318K and a novel variant of MITF gene (V320I) with risk of cancers of different site of origin in Polish population. We assayed for the presence of the E318K and V320I missense mutations in 4,226 patients with six various cancers (melanoma, kidney, lung, prostate, colon, breast) and 2,114 controls from Poland. The E318K mutation was detected in 4/2114 (0,19%) of the Polish control population, the V320I in 3/2114 (0.14%) of the controls. We found no statistically significant differences in the prevalence of the E318K and V320I among cases and controls. We found two carriers of the E318K among melanoma patients ($p = 0.95$), one carrier among breast cancer patients ($p= 0.77$), one carrier among colorectal cancer patients ($p= 0.82$) and one carrier among kidney patients ($p= 0.64$). Our study demonstrates a lack of strong association between E318K and V320I and increased risk of melanoma, cancers of the kidney, breast, prostate, lung and colon.