The effect of Mt-DNA mutations on oxidative phosphorylation in endometrial cancer genesis.

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Doctoral dissertation-abstract

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**Introduction**

Until now, there are few studies concerning mutations influence of oxidative phosphorylation system genes in mtDNA on endometrial cancer development. The present study is an attempt to complete the knowledge concerning the meaning of mtDNA disorders in endometrial cancer development and potential importance of variants *MT-CO* genes for prognosis of endometrial cancer patients.

**Aim**

The specific purpose of this study was to assessment of A5935G, G5949A, G6081A, G6267A mutations in *MT-CO1* gene and T9540C in *MT-CO3* gene, and alterations detected during the analysis of *MT-CO* gene fragments in subject and control groups. A secondary aim was to assessment of the relationship between *MT-CO1* and *MT-CO3* gene alterations and endometrial cancer incidence and evaluation of the prognostic value of *MT-CO1* and *MT-CO3* gene alterations.

**Methods**

Tested samples were taken from 100 women operated on for endometrial cancer in the Department of Gynecology ICZMP in Lodz from 2007 to 2014 (subject group). The samples were collected from 25 patients diagnosed with abnormal vaginal bleeding who underwent dilation and curettage (D&C) at the Department of Gynecology from 2011 to 2012 (control group).

Methods for the trial included: DNA isolation, polymerase chain reaction PCR, DNA sequencing. These procedures were performed according to manufacturer’s instructions.

**Results**

The T9540C mutation in *MT-CO3* gene was detected in one patient from the subject group. None of the remaining mutations were detected. The research showed that the presence of alterations in *MT-CO1* and *MT-CO3* genes typical of other types of cancer is not a risk factor for endometrial cancer.

Analysis of *MT-CO1* and *MT-CO3* gene fragments revealed 10 alterations. The alterations detected were identified in 10% of the tested group and 8% of the control group.

**Conclusion**

The research showed that the presence of alterations in *MT-CO1* gene (A5935G, G5949A, G6081A, G6267A) typical of other types of cancer is not a risk factor for endometrial cancer. Five new alterations detected in this study (C6045T, A6052G, G9548A, A9545G, G9575A) were described for the first time. Also other *MT-CO1* and *MT-CO3* mutations were detected (A6047G, T6152C, T5999C, G5913A, G9477A), previously reported in prostate cancer, hydatidiform mole, pancreatic tumor, glioblastoma multiforme, ovarian and thyroid cancers, head and neck squamous cell carcinoma.