Analysis of the association of *APOBEC3B* deletion with familial breast and/or ovarian cancer risk in Polish population.

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AID/APOBEC family consists of 11 cytidine deaminases that possess a capability of introducing sequence alterations in DNA and RNA. Proteins from AID/APOBEC family are involved in various cellular processes, including antibody diversification and innate response against retroviruses. Some of the AID/APOBEC members (primarily APOBEC3A and APOBEC3B) were also reported to be possible mutagenic enzymes responsible for induction of specific hypermutation patterns in several cancer types, including breast cancer. Recently, an association of common CNV in the *APOBEC3* cluster (deletion of the *APOBEC3B* gene) with breast cancer risk was reported in both Chinese and European populations (OR~1,3).
Our project is aimed at the characterization of the *APOBEC3B* deletion structure, development of molecular tests for the deletion genotyping and analysis of the deletion contribution to familial risk of breast and/or ovarian cancer. Firstly, we performed a sequencing analysis of the *APOBEC3B* deletion breakpoints. Based on the identified breakpoints, we designed a simple PCR-based test that allows for genotyping of the deletion in a large group of DNA samples. Additionally, we designed MLPA assay that was utilized for the independent confirmation of structure and genotypes of *APOBEC3B* deletion. We subsequently used the designed tests for the analysis of the *APOBEC3B* deletion in groups of patients with familial breast and/or ovarian cancer, patients with unselected ovarian cancer, and controls. Preliminary results confirmed association of *APOBEC3B* deletion with breast cancer and indicated that the deletion may contribute to aggregation of breast and/or ovarian cancer cases in family. Due to low frequency of *APOBEC3B* deletion in Polish population (~6%) the obtained results are only marginally significant and have to be replicated in larger groups of cases and controls.

Both PCR-based test and the designed MLPA assay will be freely available to research community and can be used to study the *APOBEC3B* deletion in other populations or other cancer types.

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