The study of genomic variation of BRCA genes in the Albanian population

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According to many studies, BRCA genes are considered polymorphic and BRCA variants are highly deleterious in damaging BRCA function, causing genome instability and increased cancer risk, affecting mostly breast and ovaries [1], [3]. This study aims to investigate the spectrum of BRCA1 and BRCA2 genetic variants in female Albanian population and to analyze the mutations significance from an evolutionary perspective. The national screening of women for breast cancer is done through imaging techniques at Obstetrics and Gynecological University Hospital (Queen Geraldina), in Tirana, Albania (almost 600 tests/year). BRCA gene variants will be identified through Multiplex Ligation Probe Assay (MLPA) and fragment analysis will be done with SeqStudio genetic analyser. These variants will be compared with the documented identified variants, to define the pathogenic variants. Actually, a family of 6 women members, already identified by MLPA assay with a deletion in exon 11 of BRCA1 gene, will be included in the positive control group. These tests demonstrated the existence of an autosomal dominant pattern of inheritance of the breast cancer cases, that is already confirmed by other studies. Furthermore, extensive studies suggest that BRCA genes vary by ethnicity [4]. This is very important to study in Albanian population, as this country has its unique genetic, linguistic, and cultural features [2]. Evolution factors contributed to this ethnic specificity, which include the positive selection imposed on human BRCA genes, the adaptation of different ethnic populations to their living environment, the bottleneck and founder effects, will also be involved in this study. As the studies suggest, this investigation is of highly importance in providing genetic evidences to understand the relationship between human evolution and cancer risk [4]. Finally, this study may contribute to provide an overview of Albanian population for the BRCA variation genes and a comprehensive reference for clinical applications in the era of precising medicine.

References

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