Genetic analysis of VEGF gene polymorphisms in women with recurrent pregnancy loss; development of a SNaPshot genotyping technique.

Email: merita.xhetani@fshn.edu.al

Vascular Endothelial Growth Factor A (VEGF gene) has significant role in angiogenesis and recently studies suggest evidence for its implications in embryo development and critical role in fetal and placental angiogenesis where formation of vascular anomalies contribute to recurrent pregnancy loss (RPL). Here we designed a multiplex single-base extension method SNaPshot for identification of variants within the VEGF gene among a group of women experiencing recurrent early pregnancy loss. All individuals were analyzed for the presence variants in the VEGF gene using a multiplex SNaPshot kit for the reaction followed by capillary electrophoresis on a SegStudio Genetic Analyzer (Applied Biosystems). Of the total of 91 women, 48 with pregnancy losses and 43 controls, 41 had more than two pregnancy losses. We excluded the cause of miscarriage by aneuploidy in the POCs (trisomi 13, 18, 21, X,Y). Two variants in the VEGF gene respectively (c.-1154G/A (rs1570360) and c.*237C/T (rs3025039) were identified and their presence observed in higher prevalence of the heterozygous and mutant homozygous genotypes for the both VEGF variants in group of women experienced RPLs compared with controls. All the variants of VEFG gene play a role in an increased frequency of RPL, however c.*237C/T genotypes seems to affect the embryo development and placental angiogenesis. By optimizing this technique, we can develop improved protocol and adding biomarkers in monitoring pregnancy outcome.

References

- [1] Macklon NS, Geraedts JP, Fauser BC. Conception to ongoing pregnancy: the 'black box' of early pregnancy loss. Human reproduction update. 2002 Jul 1;8(4):333-43
- [2] Eshre Guideline Group on RPL, Bender Atik R, Christiansen OB, Elson J, Kolte AM, Lewis S, Middeldorp S, Nelen W, Peramo B, Quenby S, Vermeulen N. ESHRE guideline: recurrent pregnancy loss. Human reproduction open. 2018;2018(2):hoy004.
- [3] Samli H, Demir BC, Ozgoz A, Atalay MA, Uncu G. Vascular endothelial growth factor gene 1154G/A, 2578C/A, 460C/T, 936C/T polymorphisms and association with recurrent pregnancy losses. Genet Mol Res. 2012; 11(4): 4739-4745.

nanoBalkan2024 Tirana (Albania)

^{1,2}Merita Xhetani, ^{2,3}Suela Leli, ¹Albina Hasa, ³Eliona Demaliaj

¹University of Tirana, Faculty of Natural Sciences, Department of Biology, Tirana, Albania

²Center of Molecular Diagnostics and Genetic Research, SUOGJ "Mbretëresha Geraldinë", Tirana, Albania

³University of Medicine and University Hospital Obstetrics-Gynaecologic SUOGJ "Mbretëresha Geraldinë", Tirana, Albania