The effect of hereditary thrombophilia on the formation of carotid artery disease: a pilot study

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Introduction: Carotid artery disease (CAD) is the narrowing of carotid arteries due to atherosclerosis. It can cause stroke. Some hereditary determinants can affect atherosclerosis formation.¹ In present study, we investigated the hereditary thrombophilia on the formation of CAD.

Patients and Methods: We evaluated the effects of Factor V LEIDEN, Factor V H1299R, Prothrombin G20210A, Factor XIII V34L, B-Fibrinogen -455 G>A, PAI-1 4G/5G, HPA1, MTHFR C677T, MTHFR A1298C, ACE I/D, APO B R3500Q, and APO E polymorphisms on CAD formation by using a ViennaLab CVD Strip Assay. Group A includes 41 patients (70.2 ± 8.6 years, 30 men) with CAD and Group B includes 39 healthy controls (67.3 ± 9.2 years, 28 men). Twenty patients had transient ischemic attack or stroke, 21 had carotid artery stenosis, more than 50 % in Group A. Hyperlipidemia is more frequent in Group A compared Group B (71%, 49 %; p<0.05). Hypertension, smoking habit and diabetes mellitus are similar in both groups.

Results and Conclusion: Heterozygote form of Factor V H1299R, Factor XIII V34L, B-Fibrinogen -455 G>A, MTHFR C677T and MTHFR A1298C were more frequent in Group A compared with Group B significantly [(2.6%, 7.3% p<0.05), (12.8%, 19.5% p<0.05), (12.8%, 19.5% p<0.05), (20.5%, 34.1% p<0.05), (25.6%, 46.3% p<0.05)]. On the formation of CAD, Factor V H1299R, Factor XIII V34L, B-Fibrinogen -455 G>A, MTHFR C677T and MTHFR A1298C heterozygous mutation seems to be determinant (p<0.05). We have some difficulty on the explication that why heterozygous form is significant even though homozygous form is not significant. This is a pilot study. We will go on working on the project to evaluate the hereditary thrombophilia.

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Zuntar I, Antoljak N, Vrkić N, Topić E, Kujundzić N, Demarin V, et al. Association of methylenetetrahydrofolate (MTHFR) and apolipoprotein E (apo E) genotypes with homocysteine, vitamin and lipid levels in carotid stenosis. Coll Antropol. 2006 Dec;30(4):871-8. PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17243563

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