



Polymorphism of rs2228145 and rs599839 as a Predictor in Development of Abdominal Aortic Aneurysm in Germany Population

Nyityasmono Tri Nugroho

University Hospital of Münster, Germany.

Abstract:

Background AAA is a degenerative vascular disease. Predominant risk factors are smoking, male, family history of AAA, and CAD. Reports indicate that two proteins are significantly up-regulated in the aortic wall of AAA, sortilin-1 responsible for circulating lipoprotein and interleukin 6 (IL-6) a pro-inflammatory interleukin. An SNP of sort1 rs599839 A>G in vicinity of gene encoding sortilin 1 and il6r rs2228145 A>C might play a significant role in the development of AAA. This study aims to identify the predictive power of those SNPs for the personalized risk assessment for developing AAA in the Germany population. Furthermore, with the determination of the single nucleotide polymorphism of selected gene, will detect the probability of AAA in the future.

Methods:

Single-centered case-control study with AAA patients diagnosed >40 mm in diameter and healthy individuals as a control were recruited after approval of ethical committee. The DNA was isolated from blood in PAXgene DNA (PreAnalytiX) with the PAXgene DNA Kit. PCR products were purified (PeqGOLD Kit, VWR) and sequenced according to Sanger (GATC Eurofins). The target sequences were analysed by SNAPGene (GSL Biotech LLC), Clustal Omega (EMBL-EBI). Statistical analysis was performed by SPSS 25 (IBM) and HaploView (Broad Institute) was used for the Linkage of Disequilibrium (LD) detection

Results:

In total 92 individuals were evaluated (56 patients). The 80.4% of patients were male, 57.1% were smoker and 16% had relatives with AAA. Mean of age, Body Mass Index (BMI), and diameter of aneurysmal of patients group were 72 ± 7.7 years, 27.1 ± 4.3 , and 56.7 ± 11.5 mm, respectively. Sort1 SNP was identified as wild homozygote in 84.8% (78/92), heterozygote in 15.2% (14/92), and rare homozygote in 0% (p value .04). The minor allele fre-



quency (MAF) for patient was .09, and for control was .06. For il6r SNP was in the same amount with sort1 SNP, nevertheless the MAF for patient was .04, and for control was .13. The LD was $D' = .08$ ($r^2 = .006$).

Biography:

Nyityasmono Tri Nugroho is a lecturer of Faculty of Medicine University of Indonesia, Jakarta, Indonesia and expert in the vascular and endovascular surgery also in genetic of vascular biology. Since 2010 he trained his general surgery and vascular surgery field then since 2016 he is pursuing his PhD in Münster, Germany, in the center of vascular and endovascular surgery also. He focused on the development of the genetic predictors for abdominal aortic aneurysm and also received many scholarship grants from Ministry of Finance, Ministry of Education Republic of Indonesia, Bank of Japan, etc. He was also invited as international speaker and many of oral and poster presentations in the vascular and endovascular surgery field such as in International Union of Phlebology (IUP) in Cortina d' Ampezzo, Italy; Indonesian Students Association in Moscow, Russia; Asian Society of Vascular Surgery Congress in Guangzhou, China; LINC Endovascular in Leipzig, Germany; also Vascular Annual Meeting in Boston, USA. He contributes a lot in development of vascular and endovascular surgery in the in Indonesia and also Asia, not only the health services, but also in educational level..

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