Povezanost genskog polimorfizma RANK/RANKL/OPG sustava s učestalošću aortne stenoze – preliminarni podaci

RANK/RANKL/OPG gene polymorphisms and aortic valve stenosis – preliminary data

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Uvod: Aortna stenoza (AS) predstavlja najučestaliji oblik degenerativne bolesti srčanih zalistaka među odraslim osobama zapadnog svijeta, sa sve većom prevalencijom zahvaljujući rapidnom starenju populacije. Trenutno ne postoje razvijeni klinički pristupi za terapiju i zaustavljanje progresije AS, a kirurška AVR (engl. *aortic valve replacement*) ili manje invazivna TAVR (engl. *transcatheter aortic valve replacement*) zamjena oštećenih aortnih zalistaka mehaničkom ili biološkom protezom predstavlja zasad jedini modalitet njenog liječenja u uznapredovalom stadiju bolesti. Brojna istraživanja pokazala su prisutnost različitih genetskih promjena kod pacijenata sa stenozom aortnih zalistaka.[1-3] Svrha ove studije je procijeniti povezanost genskog polimorfizma RANK/RANKL/OPG sustava s rizikom i kliničkim stupnjem AS. Prikazani preliminarni podaci odnose se na rs3102735 (C/T tranzicija, supstitucija) SNP polimorfizam osteoprotegrin (OPG/TNRSF11B) gena.

Pacijenti i metode: Studijom je obuhvaćeno 92 pacijenta sa AS i 131 zdravih kontrolnih ispitanika. rs3102735 SNP polimorfizam osteoprotegrin (OPG/TNRSF11B) gena ispitan je kvantitativnom lančanom reakcijom polimeraze u stvarnom vremenu (qRT-PCR) korištenjem „TaqMan® SNP Genotype Assay" kita (Life Technologies Corporation, Carlsbad, California, USA).

Rezultati: Distribucija genotipova i alela rs3102735 (C/T tranzicija, supstitucija) OPG/ TNRSF11B gena kod pacijenata s AS (CC=2.2%, CT=30.4% and TT=67.4%; C=17.4%, T=82. 6%) statistički se ne razlikuje (p>0.05) od vrijednosti dobivenih u kontrolnoj skupini ispitanika (CC=0.8%, CT=22.9% and TT=76.3%; C=12.2%, T=87.8%). Također nisu zabilježene statistički značajne razlike između pacijenata s AS i kontrolne skupine ispitanika stratificiranih s obzirom na spolu ispitanika.

Zaključak: rs3102735 SNP polimorfizam osteoprotegrin (OPG/TNRSF11B) gena nije povezan sa učestalošću AS.

Objective: Aortic valve stenosis (AS) is the most frequent heart valve disease among adults in the Western societies with ever increasing prevalence due to the rapidly ageing population. Currently there are no effective pharmacological therapies to prevent or slow the progression of AS and the surgical aortic valve replacement (AVR) or less invasive transcatheter aortic valve replacement (TAVR) procedure is still the only clinical therapy at hand for its successful treatment. Numerous research studies have shown association of genetic polimorphism with the prevalence of aortic valve stenosis.[1-3] Aims of this study are to assess the impact of RANK/RANKL/OPG gene polymorphisms on risk and severity of aortic stenosis. Herein we present the data for the rs3102735 osteoprotegrin (OPG/TNRSF11B) gene polymorphism.

Patients and Methods: The study included 92 AS patients and 131 healthy control subjects. The rs3102735 OPG gene polymorphism was identified using the quantitative real time polymerase chain reaction (qRT-PCR) and the TaqMan® SNP Genotyping Assay (Life Technologies Corporation, Carlsbad, California, USA).

Results: The OPG rs3102735 (C/T transition, substitution) genotype and allele distribution in AS patients (CC=2.2%, CT=30.4% and TT=67.4%; C=17.4%, T=82. 6%) did not significantly (p>0.05) differ from those in control group (CC=0.8%, CT=22.9% and TT=76.3%; C=12.2%, T=87.8%). Also, no statistically significant difference was found between the AS patient and control subject group stratified by gender.

Conclusion: This patient-control study shows that rs3102735 osteoprotegrin (OPG/TNRSF11B) gene polymorphism is not genetic risk factors for AS..

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