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GENETICS, GENOMICS, PROTEOMICS, METABOLOMICS

PATHOGENICITY AND LONG-TERM OUTCOMES OF LIDDLE SYNDROME CAUSED BY A NONSENSE MUTATION OF SCN11G IN A CHINESE FAMILY

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Objective: Liddle syndrome (LS) is a common monogenic hypertension with continuous activation of epithelial sodium channels (ENaCs) encoded by SCNN1A, SCNN1B, and SCNN1G. This study aimed to identify the pathogenicity of a nonsense mutation in SCNN1G in a Chinese family with LS and the outcomes of tailored treatment with amiloride.

Results: Four family members presented with severe hypertension and hypokalemia, and two had stroke. This nonsense variant resulted in a termination codon at codon 572, truncating the Pro-Pro-Pro-X-Tyr motif. The mutant epithelial sodium channels displayed higher amiloride-sensitive currents than the wild-type channels (P < 0.05). Tailored treatment with amiloride achieved ideal blood pressure control in the patients, and no adverse events occurred during follow-up.

Conclusions: We found the pathogenicity of a nonsense SCNN1G mutation (p.Glu571*) with enhanced amiloride-sensitive currents in a LS family. Tailored treatment with amiloride may be an effective strategy for the long-term control of blood pressure and the prevention of cardiovascular events in these patients.

MICRONRNA PROFILE ANALYSIS IN PERICORONARY ADIPOSE TISSUE OF DIABETIC PATIENTS WITH SIGNIFICANT CORONARY ARTERY DISEASE

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Objective: Pericoronary adipose tissue (PCAT) regulates arterial homeostasis, is considered to act in paracrine manner, and plays a role in the pathogenesis of atherosclerosis. PCAT may be a source of microRNAs (miRs) that target other tissues and act as messengers for intercellular communication. In this study, we investigated whether the PCAT surrounding coronary occlusive atherosclerotic lesions shows specific miRs expression patterns in patients with diabetes type 2 compared to non-diabetic patients.

Design and method: We enrolled 46 patients (30 men, aged 65 ± 12 years old) with 3- vessel coronary artery disease who underwent elective coronary bypass surgery with and without diabetes type 2. The PCAT samples were received from all participants. miR-133a, miR-21, miR-20b, miR-9 and miR-143 expression levels in PCAT cells were quantified by real-time reverse transcription polymerase chain reaction.

Results: Twenty-four patients with diabetic type 2 (15 men, 64 ± 10 years old) and twenty-two non-diabetic patients (15 men, 62 ± 15 years old) were included in the study. PCAT analysis showed a significant upregulation of miR-21 levels in diabetic compared to non-diabetic patients (184 ± 77 versus 21 ± 15, p = 0.04). Diabetic patients also revealed a significant increase of miR-20b and miR-143 expression in PCAT samples compared to on-diabetes (33 ± 22 versus 6 ± 13, p = 0.02, 93 ± 42 versus 16 ± 23, p = 0.01). No significant differences between the two sites were observed in PCAT expression of miR-133a and miR-9 (73 ± 12 versus 130 ± 143, 56 ± 44 versus 34 ± 33, respectively, p = NS for both).

Conclusions: miRs expression in PCAT from diabetic patients with significant coronary disease show a distinct expression profile. Our study opens new perspectives in the pathophysiological role of PCAT in atherosclerotic complications of diabetes and should be further investigated.

A STUDY OF SOME GENETIC FACTORS FOR FIBROMUSCULAR DYSPLASIA

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Objective: Fibromuscular dysplasia (FMD) is relatively rare idiopathic nonatherosclerotic noninfectious disease of the layers of the elastic arteries. Up to the current knowledge, there are not genes/mutations, which can be unequivocally associated with the disease. Eastern Europeans are under-studied and under-represented in the studies in the field. We started to study this group from the database of the Molecular Medicine center Sofia, BG and tested it to rs9349379 in gene PHACTR1, which showed some association in other studies, but is also associated with coronary artery disease (CAD) and arterial hypertension (AH).

Design and method: Eleven patients with angiographically proven FMD, 108 patients with AH, 89 patients with angiographically proven CAD and 112 healthy population controls were included. We used TaqManTM technology (Applied Biosystems) for the real time PCR sequencing of the rs9349379 in gene PHACTR1. SPSS 19, p = 0.05, CI 95% was used for the statistics.

Results: The allelic and genotypic frequencies of rs9349379 in Bulgarians (Eastern European) do not differ from the European. The frequency of the A allele was higher (without reaching statistical significance) in FMD patients. This is in accordance with the results from literature. There was also a tendency (without reaching statistical significance) for association of G allele with CAD.

Conclusions: We were not able to prove a significant association between rs9349379 in gene PHACTR1 and FMD. However, the group of FMD patients is very small. Maybe FMD is not the perfect ‘‘candidate’’ disease for genetic studies for the very few familial cases, multiple genes overlapping in common diseases, varieties in the clinical presentation. Nonetheless, this can be the basis for further research and attempts to study the disease should continue.