Title of the project: Genetic plymorphisms, MicroRNAs and bioindicators of activity: interrelations in the diagnostics and therapy of severe familial hypercholesterolemia

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Summary of 2017 results

Title of the presentation: Analysis of circulating miRNAs in patients with familial hypercholesterolaemia treated by LDL/Lp(a) apheresis

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Background: LDL/Lp(a) apheresis therapy is a well-established method of aggressively lowering LDL and Lp(a). Recently, miRNAs have been discussed as markers of vascular status including atherosclerosis. MiRNAs inhibit post-transcriptional processes through RNA duplex formation resulting in gene silencing or regulation of gene expression.

Materials and methods: We measured a profile of 175 plasma-circulating miRNAs using pre-defined Serum/Plasma Focus Human microRNA PCR Panels in pooled samples of 11 subjects with familial hypercholesterolaemia under long-term apheresis treatment. Subsequently we analysed expressions of ten pre-selected miRNAs potentially involved in lipid homeostasis in the same group of subjects. We compared plasma-circulating miRNA levels isolated from peripheral blood collected immediately before and after apheresis.

Results: The greatest differences in plasma levels were found in miR-451a, miR-16, miR-19a/b, miR-223 and miR-185. In subsequent individual miRNA assay we detected a significant increase in miR-33b levels after apheresis ($P < 0.05$). Additionally, correlations between plasma lipids and miR-33a ($P < 0.04$) and miR-122 ($P < 0.01$) have been determined. Moreover, miR-122 levels in LDLR homozygotes were higher compared to heterozygotes after, but not before, apheresis treatment ($P < 0.04$).

Conclusions: LDL/Lp(a) apheresis has an impact on miRNAs associated with lipid homeostasis and vascular status.

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