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RESEARCH AREA

Molecular genetics, functional genetics, molecular biology, lipotoxicity, lipid metabolism disorders, desaturation.

Whole-genome sequencing, now routinely used, has resulted in the identification of countless human genetic variations. However, this vast amount of information can only be interpreted in the context of a functional analysis of genetic variation. Our group is working on the molecular biological characterization of conditions associated with disorders of lipid metabolism (diabetes, obesity). Our experiments focus on the central enzymes of unsaturated fatty acid synthesis, the Stearoyl-CoA desaturases (SCD). The main function of SCDs is the on-demand channeling of saturated fatty acids towards unsaturated fatty acids, making them indispensable in many physiological processes (signal transduction, energy storage, membrane fluidity). However, their abnormal function can induce pathological processes by shifting the optimal fatty acid profile. DNA modifications in SCDs can significantly affect protein function. Genetic variations filtered by our own expression data and different prediction algorithms are characterized using classical molecular biological methods. Variants that are found to be relevant are also investigated in clinical patient samples.

TECHNIQUES AVAILABLE IN THE LAB

- genetic modification techniques: cloning, mutagenesis, PCR, RT-PCR
- investigation of regulatory and coding regions: using dual luciferase reporter systems, expression vectors, tagged constructs, fusion proteins, fluorescence microscopy or two-intron minigene vectors
- classical molecular biological methods: DNA and RNA isolation, qPCR, immunoblotting, transfection, cell culture, genotyping methods
- lipid profiling: MS, GC-FID
- mRNA and microRNA sequencing and their bioinformatic analysis, pathway analysis
- use of in silico prediction programs

SELECTED PUBLICATIONS

Orosz, G., Szabó, L., Bereti, S., Zámbó, V., Csala, M., **Kereszturi, É.** (2023) Molecular Basis of Unequal Alternative Splicing of Human SCD5 and Its Alteration by Natural Genetic Variations. Int J Mol Sci. 2023 Mar 30;24(7):6517.

Zámbó, V., Orosz, G., Szabó, L., Tibori, K., Sipeki, S., Molnár, K., Csala, M., **Kereszturi, É.** (2022) A Single Nucleotide Polymorphism (rs3811792) Affecting Human SCD5 Promoter Activity Is Associated with Diabetes Mellitus. **Genes (Basel).** 2022 Oct 3;13(10):1784.

Tibori, K., Orosz, G., Zámbó, V., Szelényi, P., Sarnyai, F., Tamási, V., Rónai, Z., Mátyási, J., Tóth, B., Csala, M., **Kereszturi, É.** (2022) Molecular Mechanisms Underlying the Elevated Expression of a Potentially Type 2 Diabetes Mellitus Associated SCD1 Variant. **Int J Mol Sci.** 2022 Jun 2;23(11):6221.

Németh, K., Tóth, B., Sarnyai, F., Koncz, A., Lenzinger, D., **Kereszturi, É.**, Visnovitz, T., Kestecher, BM., Osteikoetxea, X., Csala, M., Buzás, EI., Tamási, V. (2023) High fat diet and PCSK9 knockout modulates lipid profile of the liver and changes the expression of lipid homeostasis related genes. **Nutr Metab (Lond).** 2023 Mar 31;20(1):19.

Sarnyai, F., **Kereszturi, É.**, Szirmai, K., Mátyási, J., Al-Hag, JI., Csizmadia, T., Lőw, P., Szelényi, P., Tamási, V., Tibori, K., Zámbó, V., Tóth, B., Csala, M. (2022) Different Metabolism and Toxicity of TRANS Fatty Acids, Elaidate and Vaccenate Compared to Cis-Oleate in HepG2 Cells. **Int J Mol Sci.** 2022 Jun 30;23(13):7298.