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Preclinical development of gene therapy for the inherited child disease, creatine deficiency syndrome (CTD)

In this project we carry out the preclinical development of gene therapy methodologies for creatine transporter deficiency (CTD), an inherited childhood disease. For demonstrating safety and efficiency, we generate human induced pluripotent stem (hiPS) cell-based 3D disease model systems, then prepare two kinds of gene therapy vectors for introducing the SLC6A8 transporter into human central nervous system (CNS) progenitor cells and organoids. We generate both recombinant adeno-associated viral (rAAV) vectors and lipid nanoparticles (LNP) for brain delivery, which have already been shown to be applicable for efficient gene therapy in heritable diseases. rAAV is the current gold standard for multiple, and especially CNS-related gene therapies, some of these therapies being already in clinical applications. Since neurons do not considerably divide and proliferate, in these cells the episomal introduction of the therapeutic cDNA should provide long-term correction. LNPs are the rising stars of gene therapy, because of multiple advantages over viral gene delivery, including increased targeting possibility and less toxicity. In this project we use both transient (episomal) and stable (transposon-based) expression of the genetic constructs, providing a therapeutic correction of the missing transporter protein. Supported by LIFE_SCIENCES_I-2025-00014.

Keywords: creatine transporter deficiency, human iPS cells, brain organoid models, recombinant AAV, lipid nanoparticles

Biography

Balázs Sarkadi, MD, Ph.D., spent several years as a post-doc and then as a visiting scientist at major universities in the United States and Canada. He is research professor at Semmelweis University, member of the Hungarian Academy of Sciences, member of several international research societies including the Academia Europeae and the Royal Society of Medicine. His research has been focusing on membrane proteins in pluripotent stem cells and in stem-cell derived differentiated tissues and organoids.

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