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## RESEARCH AREA

The genome programs of the past decades have provided an enormous amount of information about the human genome and how this information is translated to the “language of life”. This knowledge is essential for understanding the pathogenesis of human diseases at the molecular level and, in fact, is currently being used to develop novel diagnostics and therapeutic modalities. Our workgroup identifies novel pathogenic mutations that result in rare monogenic human diseases. By performing functional analyses of these mutations, we attempt to understand how their mode of action leads to human disease. In another project, we investigate the genetics and molecular susceptibility factors of multifactorial human skin diseases, with a primary focus on psoriasis. We are also engaged in the investigation of non-coding RNAs. In particular, we analyze the role of the PRINS mRNA-like non-coding RNA, which was previously identified by our workgroup, in cellular stress responses and in various human diseases. In the last few years our research group has joined the Hungarian Brain Research Program (NAP Project) and as the member of the clinical branch we are engaged in the identification of genetic factors in neurodegenerative human diseases. This work has already yielded several new results for the field.

## TECHNIQUES AVAILABLE IN THE LAB

After identifying mutations using the polymerase chain reaction (PCR) and sequencing methods, various bioinformatics tools are used for sequence analysis. For our functional analyses, we employ *in vitro* DNA and cloning techniques as well as specific gene-silencing methods. Gene and protein expression is assessed using real-time reverse transcriptase PCR, western blot analysis, immunohistochemistry and immunocytochemistry. In the last few years we have introduced next generation sequencing (NGS) into our laboratory and we apply it routinely in our research work. Data provided by NGS are analyzed by various bioinformatics tools.

## SELECTED PUBLICATIONS

Tripolszki, K., Csányi, B., Nagy, D., Ratti, A., Tiloca, C., Silani, V., Kereszty, É., Török, N., Vécsei, L., Engelhardt, J.I., Klivényi, P.(5), Nagy, N., **Széll, M.** (2017) Genetic analysis of the SOD1 and C9ORF72 genes in Hungarian patients with amyotrophic lateral sclerosis. **Neurobiol Aging** **53**: 195.e1-195.e5

**Szell, M.**, Danis, J., Bata-Csorgo, Z., Kemeny, L. (2016) PRINS, a primate-specific long non-coding RNA, plays a role in the keratinocyte stress response and psoriasis pathogenesis. **Pflugers Archiv** **468**: 935-943.

**Szell, M.**, Bata-Csorgo, Z., Kemeny, L. (2008) The enigmatic world of mRNA-like ncRNAs: Their role in human evolution and in human diseases. **Semin Cancer Biol** **18**: 141-148.

Sonkoly, E., Bata-Csorgo, Z., Pivarcsi, A., Polyanka, H., Kenderessy, Szabo, A., Molnar, G., Szentpali, K., Bari, L., Megyeri, K., Mandi, Y., Dobozy, A., Kemeny, L., **Szell, M.** (2005) Identification and characterization of a novel, psoriasis susceptibility-related noncoding RNA gene, PRINS. **J Biol Chem** **280**: 24159-24167.

**Széll, M.**, Bata-Csorgo, Z., Koreckm, A., Pivarcsim, A., Polyánkam, H., Szeg, C., Gaál, M., Dobozy, A., Kemény, L. (2004) Proliferating keratinocytes are putative sources of the psoriasis susceptibility related EDA+ oncofoetal fibronectin. **J Investigat Dermatol** **123**: 537-546.