MARGIT PÁL

University of Szeged
Department of Medical Genetics

Address: Somogyi B. u. 4., H-6726 Szeged, Hungary

RESEARCH AREA

Our research group's aim is to identify the genomic causes of rare genetic disorders. Rare disease affects a small percentage of the population; its prevalence is less than 1:2000. However, taking all rare diseases together, many individuals and families are affected. To date, there are more thousands of known rare disease. They cause a large health burden to the individuals and families. 80% of rare diseases have genetic component and they are very diverse. Our research group mainly focuses on genodermatoses, neurodegenerative diseases, cardiovascular diseases, rare inheritable eye diseases, inherited hearing loss and congenital developmental abnormalities. Our findings help to understand the genetic background of rare genetic disorders and to expand human mutational databases related to human inherited diseases. This knowledge is essential for understanding the pathogenesis of human diseases at the molecular level and it can be also useful to develop novel diagnostics and therapeutic modalities. Our results may provide a good basis to develop Hungarian population-specific test panels in different inherited diseases.

TECHNIQUES AVAILABLE IN THE LAB

We apply a wide range of laboratory methods, including classical and new ones. The regularly used methods are the following: polymerase chain reaction (PCR) and sequencing methods combined with various bioinformatics tools for sequence analysis. DNA extraction from blood and tissue samples, DNA quantitation, primer design, different PCR techniques such as standard, Repeat-Primed PCR, Real-Time PCR, Digital Droplet PCR, agarose gel electrophoresis, Sanger sequencing and amplicon fragment length analysis, next generation sequencing (gene panel and whole exome sequencing) and bioinformatics analysis of these data. We use clinical and mutational databases and *in silico* variant predictions for variant interpretation and also provide genotype-phenotype comparison.

SELECTED PUBLICATIONS

Rusz, O., **Pal, M.**, Szilagyi, E., Rovo, L., Varga, Z., Tomisa, B., Fabian, G., Kovacs, L., Nagy, O., Mozes, P., Reisz, Z., Tiszlavicz, L., Deak, P., Kahan, Zs. (2017) The Expression of Checkpoint and DNA Repair Genes in Head and Neck Cancer as Possible Predictive Factors. **Pathol Oncol Res 23:** 2 pp. 253-264., 12p.

Kovács, L., Nagy, Á., **Pál, M.**, Deák, P. (2020) Usp14 is required for spermatogenesis and ubiquitin stress responses in Drosophila melanogaster. **J. Cell Sci 133:** 2.

Nagy, A., Kovacs, L., Lipinszki, Z., **Pal, M.**, Deak, P. (2018) Developmental- and tissue-specific changes of ubiquitin forms in Drosophila melanogaster. **Plos one 13:** 12.

Nagy, O., **Pal, M.**, Udvardy, A., Shirras, CAM., Boros, I., Shirras, A.D., Deak, P. (2012) lemmingA encodes the Apc11 subunit of the APC/C in Drosophila melanogaster that forms a ternary complex with the E2-C type ubiquitin conjugating enzyme, Vihar and Morula/Apc2 **Cell Div 7:** 9, 17 p.

Pal, M., Nagy, O., Menesi, D., Udvardy, A., Deak, P. (2007) Structurally Related Tpr Subunits Contribute Differently to The Function of The Anaphase-promoting Complex in Drosophila Melanogaster. **J. Cell Sci** 120: 18 pp. 3238-3248., 11 p.