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RESEARCH AREA

In our department, we carry out genetic testing of germline predisposing genes for hereditary tumors. As part of routine diagnostics, we look for heritable mutations related to the clinical phenotype. We test the genetic background of cancer syndromes possessing a high degree of heredity: most often, we carry out tests in cases of familial breast and ovarian cancer, hereditary colon tumors or neuroendocrine tumor syndromes. In addition to routine diagnostics, our research activities include the functional testing of variants with unknown effects, primarily with cDNA-level tests. We also examine the correlations of known heredity with other genetic and non-genetic factors, including those affecting tumor formation, aggressiveness, and therapy response.

TECHNIQUES AVAILABLE IN THE LAB

- DNA and RNA isolation techniques
- PCR techniques: Touch-down PCR, Long-Range PCR, Real-time PCR, RT-PCR
- Sanger sequencing technique
- NGS (next-generation sequencing): with several library preparation methods for various targets: panel enrichment (capture or PCR-based), exome sequencing, genome sequencing.
- MLPA (Multiplex Ligation-based Probe Amplification)
- cDNA studies
- Promoter methylation test
- Cell culture, techniques performed on cell lines

SELECTED PUBLICATIONS

Bozsik, A., Butz, H., Grolmusz, VK., Polgár, C., Patócs, A., Papp, J. (2023) Genome sequencing-based discovery of a novel deep intronic APC pathogenic variant causing exonization. **Eur J Hum Genet 31:** 7 pp. 841-845.

Butz, H., **Bozsik, A.**,* Grolmusz, V., Szőcs, E., Papp, J., Patócs, A. (2023) Challenging interpretation of germline TP53 variants based on the experience of a national comprehensive cancer centre. **Sci Rep 13:** 1 Paper: 14259.

Butz, H., Nagy, P., Papp, J., **Bozsik, A.**, Grolmusz, VK., Pócza, T., Oláh, E., Patócs, A. (2023) PALB2 Variants Extend the Mutational Profile of Hungarian Patients with Breast and Ovarian Cancer. **Cancers 15:** 17 Paper: 4350.

Grolmusz, VK., Nagy, P., Likó, I., Butz, H., Pócza, T., **Bozsik, A.**, Papp, J., Oláh, E., Patócs, A. (2023) A common genetic variation in GZMB may associate with cancer risk in patients with Lynch syndrome. **Front Oncol 13** Paper: 1005066.

Pálla, S., Tőke, J.,* **Bozsik, A.**,* ; Butz, H., Papp, J., Likó, I., Kuroli, E., Bánvölgyi, A., Hamar, M., Bertherat, J. et al. (2023) Whole genome sequencing resolves 10 years diagnostic odyssey in familiar myxoma. **Sci Rep 13:** 1 Paper: 14658.

Bozsik, A., Papp, J., Grolmusz, VK., Patócs, A., Oláh, E., Butz, H. (2022) Reclassification of Five BRCA1/2 Variants with Unknown Significance Using Complex Functional Study. **Cancer Res Treat 54:** 4 pp. 970-984.

Butz, H., Lövey, J.,* Szentkereszty, M., **Bozsik, A.**, Tóth, E., Patócs, A. (2022) Case Report: A Novel Pathomechanism in PEComa by the Loss of Heterozygosity of TP53. **Front Oncol 12** Paper: 849004.