

ABSTRACT

Cancers represent second the most common cause of death in the Czech Republic. The most common are breast and colorectal cancers. Identification of prognostic factors improving decision-making approaches for treatment optimization belongs to the key aims of clinical research in oncology.

Carriers of mutation in cancer-susceptibility genes represent a small but clinically important group of high-risk patients. The implementation of NGS have accelerated predisposing genes analyses. The large extent of data about the presence of variants in predisposing genes is in striking contrast to only a very limited information available about clinico-pathological characteristics of mutation carriers. Determination of the risk of tumor development in carriers of rare mutations or variants of unclear significance in genes with incomplete penetrance represent substantial drawbacks of current NGS analyses. To address these issues, we have attempted i) to introduce a unified approach to NGS analysis in breast cancer patients, ii) to characterize importance of prognostic factors in BRCA1/BRCA2 mutation carriers, and iii) to identify the cancer risks in carriers of germline mutations in the CHEK2 gene.

Colorectal cancer represents seemingly histologically homogeneous disease. However, at the molecular level it can be divided into distinct subtypes differing in prognosis. Stratification of patients with metastatic colorectal cancer on the basis of serum biochemical markers allows to identify patients suitable for radical liver resection with/without primary tumor resections. In our work, we have focused on the identification of novel biochemical markers allowing to identify patients with very poor prognosis and little benefit from combined therapy. In patients with localized colorectal carcinomas, we participated in the qualitative and quantitative evaluation of the presence of tumor infiltrating lymphocytes (Immunoscore), which is an important prognostic indicator of the risk of relapsed disease.

Keywords:

breast cancer, colorectal cancer, prognosis, estrogen receptor, TFF-3, GDF-15, Immunoscore