

Symposium Thursday 15 June - TOPICS OF LABORATORY MEDICINE IN BALKAN REGION
BIOMOLECULAR LABORATORY MARKERS IN CANCER MANAGEMENT

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The laboratory analysis is a compulsory supplement to clinical evaluation in making decisions upon diagnosis and treatment of cancer, one of the leading causes of death worldwide. The traditional biochemical laboratory protocols employed in diagnosis, prognosis and monitoring of malignant diseases include testing of soluble macromolecules as tumor markers and certain common parameters corresponding to the specific type of malignancy, but their limited reliability urges the necessity of identifying new biomarkers with higher specificity, sensitivity and predictability. Biomolecular/genetic testing is an emerging field within the scope of laboratory analysis with high clinical potential based on the assumption that revealing the genetic profile unique to each individual cancer may help predict the prognosis and select an appropriate treatment to target the changes in the specific tumor.

This work reviews the currently available genomic laboratory tests and highlights their clinical and scientific relevance. Implication of aberrant transcripts BCR-ABL and PML-RAR α for the management of leukemias, detection of genetic and epigenetic aberrations associated with familial cancer syndromes (MMR gene defects in Lynch Syndrome), pharmacogenetic assays for HER2, EGFR, ALK, KRAS, BRAF, UGT1A1 testing for selection of appropriate most efficient and least toxic treatment of particular cancer types will be addressed, along with novel, recently identified markers with anticipated clinical impact.

The rapid development of high throughput technologies shifting singleplex towards multiplex testing, such as the next generation sequencing paired with bioinformatics which enable fast and affordable sequencing of the entire genome of an individual, will inevitably empower accelerated establishment of new biomolecular markers associated with the process of cancer initiation and progression. A comparative overview of the contemporary methodologies will be presented in the lecture. Multidisciplinary efforts should be made to implement the benefits from the technological advancement in the scientific process of new biomarker identification and translation of the research results into clinical practice, in order to provide best treatment for cancer patients in accordance with the principles of the precision medicine.