Increasing significance of methylation biomarkers in clinical cancer management

Tomasz K Wojdacz
Independent Clinical Epigenetics Laboratory
Pomeranian Medical University in Szczecin
Unii Lubelskiej 1,
Szczecin, Poland.
Email: tomasz.wojdacz@pum.edu.pl

In general terms epigenetic mechanisms of gene expression regulation alter gene expression without changing primary gene sequence. There is a number of epigenetic mechanisms that orchestrate gene expression and render cell phenotype. Malfunctioning of those mechanisms leads to disease such as neoplastic transformation. In principle each of those mechanisms can be targeted by treatment or a become source of biomarkers. However, currently only assays targeting DNA methylation changes are finding significantly increasing use in clinical disease management. DNA methylation is a covalent addition of the methyl groups to cytosines within CpG dinucleotide and methylation of the promoter sequences interferes with gene transcription.

There is already substantial evidence that biomarker assays targeting disease related methylation changes can effectively be utilized as biomarkers at all stages of the clinical disease management: from risk assessment through early diagnosis and treatment personalization to post treatment surveillance. Specifically, one of the largest clinical trials have recently shown that methylation biomarker-based cancer detection in liquid biopsy allows to detect cancer in patients with remarkable specificity and sensitivity. Moreover, detectable in blood methylation of *BRCA1* gene has been proposed as biomarker of predisposition to breast and ovarian cancers. The methylation of *MGMT* gene has long been used to guide treatment of glioblastoma multiforme patients. And recently, the genome wide profiling methylation has been suggested as alternative to standard histopathology method for the classification of brain tumours by the World Health Organization.

With those developments methylation biomarkers can significantly contribute and change cancer management as well as play a significant part in personalized medicine. In my talk I will review current applications of the methylation biomarkers in clinical disease management.