The future of breast cancer screening

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The main families of new biomarkers rising hopes in the very early detection of breast cancers are genomics, transcriptomics, proteomics and metabolomics.

Aberrant hypermethylation of the promoter region of tumour-suppressor genes, leading to a loss of function of the gene, is a frequent and early event in cancer cells. Sensitive DNA-methylation-specific PCR technology permits the detection of gene methylation from rare tumor cells in tissue biopsies, blood, urine, and other body fluids, with a high specificity. In the future, it would be possible to simultaneously detect a very small breast cancer and predict the future behavior of this tumor by screening with gene methylation.

Proteins are ultimately responsible for the disease phenotype. The use of highly sensitive and specific proteomics techniques coupled with the new computational capabilities permit an unbiased cataloging of molecular changes associated with cancer initiation and progression.

The ultimate goal is to be able to combine both genomics and proteomics-based approaches to the screening, discovery and validation of biomarkers of breast cancer.

However, challenges to the clinical implementation of these techniques include the need for validation in larger, well-defined populations with optimized and standardized methodology. Above all, it is mandatory to avoid over-diagnosis and over-treatment: development of new powerful prognosis factors, imaging technologies and targeted therapies have to be first achieved before to use molecular screening of breast cancer in clinical practice.