Human Genet Embryol 2016, 6:3(Suppl)
http://dx.doi.org/10.4172/2161-0436.C1.003

Micronucleus frequency and *BMI-1* expression analysis as two possible cytogenetic and molecular metastasis biomarkers in breast cancer

Mahdieh Salimi, Elaheh Eskandari and Sepehr Sarraf Zadeh
National Institute of Genetic Engineering and Biotechnology, Iran

Breast cancer is a highly invasive and metastatic disease and known for its propensity to recur decades after treatment. Breast cancer cells that undergo epithelial-to-mesenchymal transition (EMT) obtain malignant characteristic, however, the molecular mechanism and/or cytogenetic characteristic underlying this transition are poorly understood. Breast cancer patients have a reduced DNA repair capacity. B-cell-specific Moloney murine leukemia virus integration site 1 (*BMI-1*) is a Polycomb group protein that is able to induce telomerase activity, enabling the immortalization of epithelial cells. Immortalized cells are more susceptible to double-strand breaks, which are subsequently repaired by homologous recombination. Overexpression of *BMI-1* has been reported in various human cancers and proved to be associated with poor survival. Spontaneous and bleomycin-induced genomic instability and *BMI-1* gene expression in peripheral blood of breast cancer patients compared with first degree relatives (FDR) and normal control were examined using the cytokinesis block micronuclei-cytome (CBMN cyt) and real-time RT-PCR assays, respectively. Possible association between these events and a series of clinical-pathological parameters of the tumors were studied. In spite of MN frequency with no association with lymph node involvement, the peripheral blood net frequency of micronuclei and *BMI-1* expression were significantly higher in breast cancer patients with distant metastasis. *BMI-1* expression level was higher in lymph node positive (LN+) breast cancer compared with LN negatives. Our results indicate that increased genomic instability expressed as micronuclei and higher *BMI-1* expression in peripheral blood are associated with metastasis in breast cancer. Therefore implementation of micronucleus assay and *BMI-1* expression analysis in blood as possible cytogenetic and molecular biomarkers in clinical level may potentially enhance the quality of management of patients with breast cancer.

salimi@nigeb.ac.ir

Rapid copy number typing of the human glycophorin A-B-E cluster

Walid Algady and Edward J Hollox
University of Leicester, UK

Human glycophorins A and B are proteins expressed on the surface of erythrocytes and are receptors for *Plasmodium falciparum* invasion of this cell type. Both proteins are encoded by the genes *GYPA* and *GYPB* which, together with *GYPE*, reside on a tandemly-duplicated repeat region on chromosome 4q31.21. Recent genetic analysis has suggested that a particular haplotype within or close by this gene cluster is protective against severe malaria. The relation between this haplotype and copy number variation (CNV) of the glycophorin genes and the role of glycophorion gene CNV in malaria remains unclear. Here, we present a rapid, cost-effective, robust suite of assays to type copy number of this gene cluster and some preliminary data using these assays. We use the paralogue ratio test (PRT), a comparative PCR method which has been shown to be more robust than real-time quantitative PCR for typing genomic CNV and has been previously used on a wide variety of CNV loci. It requires <50 ng genomic DNA and has a low cost per sample, making it ideal for genotyping large DNA cohorts to test for association with malaria severity, for example.

wa79@leicester.ac.uk