Applications of pharmacogenetics; our experience the last six years

Ass Professor Dr Andreas Hadjisavvas
Cyprus School of Molecular Medicine-The Cyprus Institute of Neurology and Genetics

Pharmacogenetics, the science of how an individual’s genetic makeup influences the response to certain drugs, has improved significantly the decision-making process by providing critical insights into how a patient will react to a specific treatment. During the past few years, new medical treatments known as “targeted therapies” have become available. These have changed dramatically the repertoire of treatment options of non-small-cell lung cancer (NSCLC), colorectal cancer, metastatic melanoma as well as ovarian cancer.

For example results from recent clinical trials show that patients with epidermal EGFR mutation-positive NSCLC, might have a survival advantage over those with EGFR mutation-negative disease. Currently patients diagnosed with lung cancer are better stratified using an integrated approach that includes data from histology, immunohistology and molecular biology. In this context the status for the EGFR status is mandatory, for selecting the more efficacious front line therapy.

Since January 2011, pharmacogenetic tests have become an integral part of contemporary oncology practice in Cyprus. The Department of EM / Molecular Pathology of the Cyprus Institute of Neurology and Genetics is offering genetic testing by examining DNA from tumor biopsies for the EGFR and BRAF genes of lung cancer, BRAF gene for metastatic melanoma, BRAF, KRAS and NRAS genes for colorectal cancer and BRCA1 and BRCA2 genes for ovarian cancer. Over the last six years more than 1200 tumor biopsies have been tested, for the above genes, by the EM/Molecular pathology Department.