Integrating genetic information with breast cancer risk factors may help refine prognosis

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Incorporating genetic information known as gene expression signatures with clinical and other risk factors for breast cancer may help refine estimates of relapse-free survival and predicted response to chemotherapy, according to a study in the April 2 issue of JAMA.

“The advent of genomic technology for the analysis of human tumor samples has now added an additional source of information to aid prognosis and clinical decisions. In particular, the development of genomic profiles that accurately assess risk of recurrence offers the hope that this information will more precisely define clinical outcomes in breast cancer. The dimension and complexity of such data provide an opportunity to uncover clinically valid trends that can distinguish subtle phenotypes [physical manifestations] in ways that traditional methods cannot,” the authors write. Few studies have examined the value in integrating genomic information with the traditional clinical risk factors to provide a more detailed assessment of clinical risk and an improved prediction of response to therapy.

Chaitanya R. Acharya, M.S., of the Duke Institute for Genome Sciences and Policy, Duke University, Durham, N.C., and colleagues conducted a study to determine the value in incorporating genomic information with clinical and pathological risk factors to refine prognosis and to improve therapeutic strategies for early stage breast cancer. The study included patients with early stage breast cancer who were candidates for supplemental chemotherapy; 964 breast tumor samples were used. All patients were assigned relapse risk scores based on their respective

clinicopathological features. Genetic testing was performed and gene expression signatures (characteristic profiles) were applied to these samples to obtain patterns of deregulation that correspond with relapse risk scores to refine prognosis with the clinicopathological prognostic model alone. Predictors of chemotherapeutic response were also applied to further characterize clinically relevant heterogeneity (diversity) in early stage breast cancer.

The researchers found that integrating gene expression signatures into clinical risk stratification could refine prognosis for patients in three risk subgroups (low, intermediate, and high) and help predict relapse-free survival and response to chemotherapy.

“Pending future prospective clinical validation, these results provide preliminary evidence that the profusion of gene expression signatures in defining breast cancer, if used appropriately, represent less of a paradox and should be viewed as an important complementary approach to current clinicopathological risk stratification systems. Furthermore, knowledge of increased likelihood of sensitivity to specific chemotherapeutic agents from a repertoire of drugs that are commonly used to treat breast cancer is something that could be more immediately used in current clinical practice, once issues regarding cost and accessibility are addressed, in instances wherein multiple chemotherapeutics or chemotherapeutic combinations are Food and Drug Administration approved, as in early stage breast cancer, and are considered the standard of care,” the authors conclude.

Source: JAMA and Archives Journals

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