

Editorial Article

Molecular Diagnostics in Cancer Diagnosis and Management

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Editorial

Oncology has undergone remarkable changes in the past decades and has entered the era of precise medicine and personalized treatment. Cancer management is increasingly dependent on accurate diagnosis and cancer prevention. Although cancer diagnosis still largely relies on pathology, medical imaging, and other tests, molecular diagnostics is expanding in this territory. In this Editorial, we will briefly discuss the role of molecular diagnostics in cancer diagnosis and management.

Cancer is essentially a disease of the genome. Alterations in oncogenes, tumor-suppress or genes and stability genes are responsible for tumorigenesis. So far, about 300 cancer genes have been reported, constituting more than 1% of all the genes in the human genome. About 90% of cancer genes show somatic mutations in cancer, 20% show germline mutations and 10% show both [1]. Molecular diagnostics is a collection of techniques that analyze genetic materials, proteins, or related molecules and thus provide information about health and disease. It is a fundamental component of precision medicine. In cancer diagnosis and therapy, molecular diagnostics contributes in the following ways: to determine whether a person is at risk for a certain type of cancer; to help diagnose cancers or differentiate cancers from benign tumors; to determine blood cancer subtypes; to help predict whether patients will respond to cancer treatments; to evaluate the likelihood that an existing cancer will recur after treatment.

In personalized therapy for advanced non-small cell lung cancer (NSCLC), for example, molecular analyses play a vital role. Patients with sensitizing mutations in the epidermal growth factor receptor (*EGFR*) experience improved progress-free survival and response rates with first-line gefitinib or erlotinib therapy relative to traditional platinum-based chemotherapy, while patients with *EGFR*-mutation negative tumors gain greater benefit from platinum-based chemotherapy [2]. Anaplastic lymphoma kinase (*ALK*) disorders are shown to cause about 5% of NSCLC, therefore, *ALK* tyrosine kinase receptors are therapeutic targets in NSCLC and other malignancies with *ALK* fusion [3]. Molecular diagnostics can be repeated over the course of treatment in order to monitor treatment response and signs of recurrence. In *ALK*-positive patients, molecular diagnostics can identify those who might develop resistance to initial *ALK* inhibitor (e.g., crizotinib) treatment, and to whom new generation inhibitors (e.g., ceritinib and alectinib) should be administered accordingly. The College of American Pathologists, the International Association for the Study of Lung Cancer, and the Association for Molecular Pathology have published a guideline for selection of lung

cancer patients for EGFR and ALK tyrosine kinase inhibitors [4]. This guideline makes recommendations on testing for *EGFR* mutations and *ALK* fusions to guide patient selection for therapy, and is endorsed by the American Society of Clinical Oncology [5].

DNA sequencing is one of the most important molecular diagnostics technologies. Sanger sequencing, developed by Edward Sanger [6], was considered the gold standard for nucleic acid sequencing for nearly three decades. Although incremental improvements reduced the cost of Sanger sequencing and allowed laboratories to add contents to their tests, it is the massive parallel sequencing technique that facilitates high throughput sequencing [7]. This second generation sequencing, or next generation sequencing (NGS), allows whole genome or several genomes to be sequenced in a short time period. Some of the NGS technologies emerged in the 1990's and became commercially available since 2005. The past decade witnessed a revolution in sequencing technologies that is considered one of the most significant technological advances in the biological sciences in the recent years [8]. Targeted DNA enrichment methods allow even higher genome throughput at a reduced cost per sample. NGS is implemented by many laboratories for routine diagnostics, and the application of NGS in cancer leads to tremendous advances in cancer diagnosis and personalized therapy.

Call for Submissions

Greetings from the *Journal of Comprehensive Cancer Research*! It is evident from the above review that, cancer therapy has advanced far beyond the preliminary pathology-therapy model. The added complexity by molecular diagnostics and other disciplines has greatly changed cancer diagnosis and management. Contemporary cancer treatments are becoming increasingly personalized, and a greater emphasis on multidisciplinary thinking tends to make individual clinicians and researchers multidisciplinary as well. JCCR is created to serve the communities of cancer therapy, cancer research, and anti-cancer drug development. JCCR is an open access journal that is intended to provide extensive coverage in all areas of cancer. We sincerely hope that JCCR will become an invaluable platform for worldwide researchers and clinicians to exchange their findings and express their opinions in cancer research and treatment. We wholeheartedly welcome established researchers as well as young investigators. Authors may find a variety of manuscript types with which to report interesting studies: original research paper, review article, mini-review, case report, commentary, short communication, and letter to the editor. As the Journal grows, the latest research and clinical findings will rapidly disseminate, and thus the authors and readers of JCCR will mutually benefit.

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