The Evolving Role of Genetic Testing in Cancer Treatment: Current Status and Future Perspectives

Yoshiki Shinomiya
Center for Development of Advanced Diagnostics, Hokkaido University Hospital

Molecular targeted therapy plays a crucial role in cancer treatment by specifically targeting abnormal proteins that drive cancer development and progression. By focusing on these molecularly altered proteins produced by genetic mutations, targeted therapies can minimize damage to normal cells while effectively inhibiting tumor growth and metastasis. Therefore, molecular targeted therapy is an important cancer treatment option.

To effectively implement molecular targeted therapy, it is essential to accurately identify genetic mutations in the cancer genome that can serve as therapeutic targets. This process is often performed as part of a companion diagnostic test (CDx). Traditionally, genetic mutation detection has relied on single-gene testing methods, such as immunohistochemistry, fluorescence in situ hybridization, and the polymerase chain reaction. However, as the genetic diversity of cancers has become increasingly evident, the number of molecular targets has increased, leading to the development of a broader range of targeted therapies. In response to this, next-generation sequencing (NGS)-based comprehensive genetic testing has emerged as the preferred approach for detecting mutations.

CDx is most frequently performed for non-small cell lung cancer (NSCLC). It is known that driver mutations in the *EGFR*, *KRAS* (G12C), *ALK*, *RET*, *MET*, *ROS1*, *BRAF* (V600E), *ERBB2* and *NTRK* genes occur in a mutually exclusive manner in NSCLC, with studies indicating that approximately 50% of patients harbor one of these mutations. Given this landscape, the use of NGS-based panel testing has increased, allowing the simultaneous detection of multiple actionable mutations.

Moreover, comprehensive genomic profiling (CGP) tests such as the NCC Oncopanel System, FoundationOne® CDx, and GenMineTOP™ have gained prominence. These tests facilitate the broad analysis of hundreds of cancer-related genes by detecting mutations, amplifications, deletions, and fusions. Additionally, some CGP tests assess biomarkers such as microsatellite instability (MSI) and tumor mutation burden (TMB), which are relevant to the efficacy of immune checkpoint inhibitors. Notably, patients with MSI-high or TMB-high tumors show a favorable response to immune checkpoint inhibitors, thereby expanding their therapeutic options.

However, CGP tests can also detect alterations of uncertain clinical significance. For this reason, test results are usually reviewed by multidisciplinary expert panels comprising oncologists, clinical geneticists, pathologists, and genomics specialists to determine the most appropriate treatment strategies for each patient. Furthermore, these data are stored at the Cancer Genome Information Management Center of Japan's National Cancer Center and can be utilized for research by academic institutions and pharmaceutical companies, with patient consent. This data-sharing framework not only informs current clinical practice but also contributes to the development of future therapies.

Currently, only approximately 10% of patients who have undergone CGP testing receive treatment based on the results. However, with the rapid progress in targeted therapy research, this percentage is expected to increase. Additionally, national initiatives to introduce whole-genome sequencing into clinical practice are underway, paving the way for a more comprehensive approach to cancer treatment. The continued expansion of genetic testing will enable increasingly precise and personalized therapeutic strategies, providing more optimal treatment options to patients with cancer.

In this presentation, I provide an overview of the current landscape of genetic testing and targeted therapies in cancer treatment and explore future directions in this rapidly evolving field.

癌治療における遺伝子検査の現状と今後

四宮　義貴
北海道大学病院 先端診断技術開発センター

分子標的治療は、がんの発生と進行を促す異常なタンパク質を標的とすることで、がん治療において重要な役割を果たす。遺伝子変異によって生成された分子レベルで変化したタンパク質に焦点を当てることで、標的治療は正常な細胞へのダメージを最小限に抑えながら、腫瘍の成長と転移を効果的に抑制することができる。そのため、分子標的治療は重要ながん治療の選択肢となっている。

分子標的療法を効果的に実施するには、治療の標的となり得る癌ゲノムにおける遺伝子変異を正確に特定することが不可欠である。このプロセスは、コンパニオン診断（CDx）の一環として実施されることが多い。従来、遺伝子変異の検出には免疫組織化学、蛍光 in situ ハイブリダイゼーション、ポリメラーゼ連鎖反応などの単一遺伝子検査法が用いられてきた。しかし、がんの遺伝的多様性が明らかになるにつれ、分子標的の数も増加し、より幅広い標的療法の開発につながった。これを受けて、変異の検出には次世代シーケンシング（NGS）に基づく包括的遺伝子検査が最適なアプローチとして登場した。

CDxは非小細胞肺がん（NSCLC）に対して最も頻繁に行われている。NSCLCでは、*EGFR*、*KRAS*（G12C）、*ALK*、*RET*、*MET*、*ROS1*、*BRAF*（V600E）、*ERBB2*、*NTRK*遺伝子におけるドライバー変異が相互に排他的に発生することが知られており、これらの変異の1つを保有する患者は約 50%であることが研究で示されている。このような状況を踏まえ、NGS ベースのパネル検査の利用が増え、複数の治療可能な変異を同時に検出できるようになった。

さらに、NCC Oncopanel システム、FoundationOne® CDx、GenMineTOP™ などの包括的ゲノムプロファイリング（CGP）検査が注目されるようになった。これらの検査では、変異、増幅、欠失、融合を検出することで、数百種類のがん関連遺伝子を幅広く解析することが可能である。さらに、CGP 検査の中には、免疫チェックポイント阻害剤の有効性に関連するマイクロサテライト不安定性（MSI）や腫瘍変異負荷（TMB）などのバイオマーカーを評価するものもある。注目すべきは、MSI-high または TMB-high の腫瘍を持つ患者は免疫チェックポイント阻害剤に良好な反応を示すため、治療の選択肢が広がる点である。

しかし、CGP 検査では臨床的意義が不明確な変異も検出されることがある。このため、通常は腫瘍内科医、臨床遺伝専門医、病理医、ゲノム医などの専門家による多職種の検討会で検査結果を精査し、患者一人ひとりに最適な治療方針を決定する。また、これらのデータは国立がん研究センターがんゲノム情報管理センターに保管され、患者の同意を得た上で、学術機関や製薬企業による研究に活用されている。このデータ共有の枠組みは、現在の臨床に役立つだけでなく、将来の治療法の開発にも貢献する。

現在、CGP 検査を受けた患者のうち、検査結果に基づく治療を受けている患者は約 10%にとどまっている。しかし、標的療法の研究が急速に進展していることから、この割合は今後増加すると考えられる。また、全ゲノムシークエンシングを臨床に導入する国家的な取り組みも進行中であり、がん治療のより包括的なアプローチへの道が開かれつつある。遺伝子検査の継続的な拡大により、より精密で個別化された治療戦略が可能となり、がん患者により最適な治療オプションを提供できるようになる。

本発表では、がん治療における遺伝子検査と標的療法の現状について概説し、急速に進化するこの分野の今後の方向性について考察したい。

The Role of Biomedical Laboratory Scientists in the Era of Genomic Medicine
Practical Application and Future Prospects of Clinical Microbial Genetic Testing Kits

MITSURU MATSUMURA
Department of Clinical Laboratory Science, Teikyo University Faculty of Medical Technology

【Background and Objectives】
Recent advances in genomic medicine have spurred significant innovations in testing technologies, steadily advancing the realization of precision medicine. Genetic testing kits based on PCR, real-time PCR, and multiplex methods are increasingly utilized in microbiology laboratories. Although next-generation sequencing (NGS) technology is still predominantly in the research phase, it is fundamentally transforming conventional testing methods by enhancing diagnostic accuracy and speed. Furthermore, a comparison of traditional microbial testing methods with genetic testing is reshaping the roles and skill sets required by biomedical laboratory scientists.

【Table of Contents】
1. Overview of Major Clinical Microbial Genetic Testing Kits and NGS Technology in Japan
This section outlines the fundamental principles and advantages of PCR-based genetic testing kits (e.g., GeneXpert MTB/RIF) that are widely employed in Japanese clinical settings. It also details the basic principles behind NGS-based testing methods and examines the technological innovations that differentiate them from conventional approaches.

2. New Roles and Necessary Skills for Biomedical Laboratory Scientists
The advent of advanced technologies has expanded traditional testing responsibilities to include the data analysis of PCR results, clinical interpretation of findings, and the dissemination of information through collaboration with physicians and other healthcare professionals. In this context, we discuss the current state of practice, emphasizing the need to enhance education and training, revamp quality management systems, and further strengthen interprofessional cooperation.

【Discussion and Summary】
Recent advancements in genetic testing kits have the potential to address a wide range of clinical challenges, such as the rapid identification of viruses, difficult-to-culture protozoa, and slow-growing bacteria. This capability facilitates prompt treatment decisions and is expected to improve patient outcomes. Concurrently, biomedical laboratory scientists are increasingly tasked with advanced data analysis, enhanced communication with clinical staff, and stringent quality control throughout the testing process. By leveraging both established testing systems and innovative technologies, these professionals are well positioned to improve diagnostic speed and precision, thereby optimizing treatment plans through effective collaboration with the medical team. This presentation explores the practical application of conventional technologies, the evolution of professional roles, and future directions for education and training.

ゲノム医療時代における臨床検査技師の役割
～臨床微生物遺伝子検査キットの実践とこれから～

松村　充
帝京大学 医療技術学部 臨床検査学科

【背景・目的】
近年、ゲノム医療の急速な発展に伴い、各種検査技術が飛躍的に革新され、精密医療の実現が着実に進展している。特に、微生物検査室で広く運用されている PCR やリアルタイム PCR、マルチプレックス法に基づく遺伝子検査キットが汎用されつつある。研究段階ながら注目を集める次世代シーケンシング（NGS）技術は、従来の検査手法を根本から変革し、診断の正確性と迅速性の向上に寄与している。また、微生物検査の従来法と遺伝子検査を比較し、臨床検査技師には新たな役割とスキルが求められている。

【内容】
1．日本における主要な臨床微生物遺伝子検査キットおよび NGS 技術の概説
　日本の臨床現場で広く採用されている PCR ベースの遺伝子検査キット（例：GeneXpert MTB/RIF 等）について、その基本原理、利点などを説明する。また、NGS 技術を用いた検査手法の基本原理と、従来法との比較から見える技術革新の背景について概説する。
2．臨床検査技師の新たな役割と必要なスキル
　最新技術の導入により、従来の単なる検査実施業務に加え、PCR 検査結果のデータ解析、結果の臨床的解釈、および医師や他職種との連携を通じた情報提供が求められる。これに伴い、教育・研修の充実、品質管理体制の再構築、さらに多職種連携の強化が不可欠となる。

【考察・結論】
　最新の遺伝子検査キットは、培養困難なウイルスや原虫、発育の遅い菌種の同定など、多様な臨床課題に対応している。これにより、臨床現場では検査結果を基に迅速な治療方針の決定が可能となり、患者アウトカムの改善が期待される。同時に、臨床検査技師には、従来以上に高度なデータ解析能力、臨床スタッフとの円滑なコミュニケーション能力、および検査プロセス全体の品質管理が求められている。
　ゲノム医療時代において、日本で広く活用される遺伝子検査キットおよび将来的な NGS 技術は、薬剤耐性菌の検出を含む多角的な臨床検査の可能性を拡大している。臨床検査技師は、これらの確立された検査システムと革新的技術を効果的に活用することで、診断の迅速化・正確化および医療チーム内での情報共有を通じた治療方針の最適化に寄与することが求められる。本発表では、従来技術の有効活用とそれに基づく役割変革の方向性、並びに今後の教育・研修の在り方について議論する。

The medical technologists’ role as a genetic counselor

The medical technologists’ role as a genetic counselor

Haeyong Jung
Soonchunhyang University Seoul Hospital

As the human genome was revealed through the Human Genome Project completed in 2003, and genetic analysis technology has recently developed through the development of next-generation sequencing methods, genetic testing has been actively implemented, leading to an increasing number of diagnoses of rare genetic diseases that were previously undiscovered. According to the World Health Organization, there are more than 6,000 types of genetic diseases. According to data from 1998, the number of people with genetic diseases in Korea was about 20,000, but in 2012, the number of patients with rare genetic diseases related to genetic mutations is estimated to be less than 1 million.

Medical technologists are professionals who can legally test human samples and have also completed sufficient genetic training, and are currently performing their role as examiners of tests that reveal rare genetic diseases. However, in the future, it is expected that the role of medical technologists will serve not only as testers for genetic diseases but also as genetic counselors.

Rare genetic diseases require knowledge about the disease and specialized knowledge about its treatment. Since there are many diseases for which treatments have not yet been developed, patients or their families need not only professional knowledge but also psychological management. They also need counseling to help them receive welfare services. However, in Korea, most genetic counseling is performed by medical doctors. Therefore, information on medical knowledge is provided.

In the United States and Japan, genetic counselors are already performing a role that includes not only professional knowledge for patients but also psychological support for patients' families. Accordingly, in Korea, it is believed that if medical technologists who have knowledge of rare genetic diseases and perform genetic audits play the role of genetic counselors, they will be able to contribute to future precision medicine and personalized medicine.

Perspectives of Clinical Laboratory Scientists on Genetic Testing
Assessing Awareness, Understanding, and Challenges in Expanding the Role of Clinical Laboratory Scientists in Health Screening Centers within Genomic Medicine

Sejin Lee
KAHP(Korea Association of Health Promotion)

In the era of genomic medicine, genetic testing, which is essential for personalized care, continues to expand in market size. Furthermore, as genetic testing becomes increasingly integrated into routine health checkups, it is also being offered at health screening centers, creating an environment where more people can access it without significant burden. Our institution also provides genetic testing through its health screening center, and the clinical laboratory scientists working here perform various tasks in addition to conducting genetic tests. This study aims to investigate the awareness, understanding, and future perspectives of clinical laboratory scientists, who carry out these diverse tasks, using a Likert scale survey. By analyzing the differences between the survey items, this study seeks to explore the potential for expanding the roles of clinical laboratory scientists.

The Role of Biomedical Laboratory Scientists in the Era of Genomic Medicine
Empowering Healthcare through Genetic Expertise and Collaborative Practice

Huang, Chung-Guei
[1]Vice President, Taiwan Association of Medical Technologist; [2]Director, Department of Medical Laboratory, Lin-Kou Chang-Gung Memorial Hospital, Chang-Gung Medical Foundation; [3]Associate Professor, Chang-Gung University

In the era of genomic medicine, the role of biomedical laboratory scientists (BMLS) is becoming increasingly pivotal in advancing healthcare and improving patient outcomes. Their essential contributions within the healthcare system span a wide range of responsibilities, including specialized expertise in genetic testing, data interpretation, and clinical collaboration. With the rapid advancement of genomic technologies, BMLS are entrusted with ensuring the accuracy and reliability of genomic analyses, which are critical for the effective implementation of personalized medicine.Moreover, BMLS play a significant role in educating other healthcare professionals and patients about the implications of genetic information, fostering a greater understanding of how genomics can influence treatment decisions and health management. By facilitating collaboration among multidisciplinary teams, BMLS help to integrate genomic insights into clinical practice, ensuring that genetic data informs patient care at every stage. This speech will highlight the ongoing need for continual professional development and adaptability among BMLS to navigate the complexities of genomic medicine. The commitment to advancing knowledge and skills is essential for coping with the rapid changes in this dynamic field. Ultimately, by reinforcing the integral role in enhancing healthcare delivery, we aim to inspire innovative approaches and underscore the importance of BMLS in shaping the future of genomic medicine. Together, we can build a healthcare system that fully leverages the power of genomics to improve patient outcomes and promote public health.