



# Knowledge-based diagnosis and prediction using big data and deep learning in precision medicine

## SMART MEDICINE: HEALTH CARE TAILORED JUST FOR YOU

By now, many of us have probably heard about (or work with) Watson for Oncology. This artificial intelligence program was developed by IBM Co. (Armonk, NY, USA) and can provide optimal and individualized treatment options for patients through its vast medical database and the patient's own unique medical and genetic information. Worldwide, Watson has already been introduced and used for actual diagnoses at several clinical centers. As evidenced by this remarkable program, we can see now that the future of medicine is near and will be mediated through the integration of technologies across multiple disciplines, such as genetics, genomics, big data, and deep learning.

Thanks to the Human Genome Project, which was launched in 1987, human genes and DNA sequences have been fully mapped out. Like unique fingerprints, human genes can vary distinguishably among individuals. Thus, we can monitor each person's health by understanding and characterizing his or her genes. Primary physicians can classify the genetic information of each patient and provide advice regarding prevention or predict diseases that have a greater probability of occurring. When the disease appears, physicians can also effectively provide tailored medications and therapies, ensuring that the most optional treatment is given to the right patient in the right dose at the right time.

To reap the benefits that genetic information can yield, it is essential to integrate big data, deep training, and bioinformatics, all of which contribute to making the connection between specific diseases and signatures. In addition, smart medicine can contribute to monitoring the health conditions of individuals. For instance, Google Health and Microsoft Blot facilitate direct and real-time communication between health care providers and patients through the use of smart phones. This has resulted in

increased active, direct, and cooperative patient involvement in decision-making processes. These advances have even given smart medical devices and software the nickname of "pocket doc." Beyond just Watson and smart medicine, progress in robotics, stem cell therapy, bio ink, and artificial tissue construction using 3-dimensional printing technology will change our future medical environment and culture.

## SMART MEDICINE IS PRECISION MEDICINE

What exactly is "precision medicine"? Why is it so promising and what can it do? Until now, most medical treatments have been designed for the average patient by using a one-size-fits-all model. This has led to partial benefits for some but not for others. Precision medicine is an innovative approach that takes into account each individual's different characteristics, such as genetic profile, environment, and lifestyle. To capture the entire spectrum of potential differences, a multi-omics approach has been considered. "Omics" has far-reaching capabilities and includes many different fields, such as genomics, transcriptomics, epigenomics, proteomics, glycomics, metabolomics, and lipidomics. Such a varied array of research avenues is believed to propel our understanding of diseases, leading to powerful new discoveries and treatments that can be tailored to individuals. It also gives medical professionals the resources needed to design specific treatments for individuals.

In 2015, the United States (US) National Institutes of Health (NIH) initiated the Precision Medicine Initiative and invested over US dollars 200 million to accelerate biomedical research and provide clinicians with new tools for selecting personalized therapies. In particular, efforts on precision oncology by the US National Cancer Institute (NCI) have focused on identifying genetic alterations that drive cancer growth. The NIH and the NCI have established a series of

precision medicine clinical trials (e.g., the NCI molecular profiling-based assignment of cancer therapy [NCI-MPACT] and molecular analysis for therapy choice [NCI-MATCH]). Furthermore, a national database for integrating cancer genomic information with clinical outcome information has also been built.

Now, this precision oncology approach has become available for patients. Routine molecular testing is becoming part of the standard of care, enabling physicians to choose the treatment that will best improve a patients' chances of survival and reduce side effects. To facilitate the successful implementation of precision oncology in routine clinical practice, several considerations should be taken into account. These include selecting the right molecular test, diverse testing options, affordable cost, quality of biospecimens, reasonable turnaround time, user-friendly bioinformatics analysis, and accurate clinical interpretation by health care providers. The infrastructure and standard operating procedures for molecular testing should be well established before moving this work forward; this also requires close communication between oncologists, pathologists, hospital administrators, clinical facilities, and laboratories. Sequencing-based clinical precision oncology programs generally prefer targeted gene panels designed for solid tumors (e.g., the Memorial Sloan Kettering Cancer Center-integrated mutation profiling of actionable cancer targets [MSK-IMPACT] or the University of Michigan's MI\_Oncoseq). Current efforts have been concentrated on maximizing the throughput, cost-effectiveness, and depth of coverage of detecting gene mutations or other critical cancer-associated genetic aberrations. PrecisionFDA of the US Food and Drug Administration has also provided a user-friendly workspace to conduct further precision oncology studies.

## USING SOCIAL MEDIA TO GATHER BIG DATA IN PRECISION MEDICINE

Health-associated big data can be obtained not only from the molecular profiles of clinical tests, but also from epidemiological data collected through social media platforms and health-related social networking sites (e.g., Facebook, Twitter, Google+, Patients Like Me, e-forums). More and more people have been using social media to share personal information about their health conditions and seek information regarding available clinical trials, educational materials, and support groups. As a new data source for research, social media has been used to identify patients, recruit for clinical trials, track patient conditions, monitor patient care, and start new discussions about

patient populations without geographical or health policy restrictions.

## ARE WE READY? EDUCATION AND TRAINING PLANS IN THE ERA OF PRECISION MEDICINE

The era of high-throughput technologies promises to accelerate our scale dramatically. Multi-omics-based molecular profiles offer an exciting opportunity for precise diagnosis and tailored treatment. However, we should also be aware of the possible overpromises and distortion of priorities in precision medicine. How can we make smart "omics" accessible to clinicians? Do clinicians have access to the necessary support for interpreting and translating this information into therapeutic interventions? Sadly, the answer seems to be "not yet." Currently, developed molecular biomarker testing for precision oncology aims to provide the best therapeutic opportunities and avoid ineffective or toxic therapies for certain patients. Unfortunately, another challenge lies in the complicated and (still) technically oriented interpretations of big data into clinically meaningful information. In addition, communication of test results to patients is another challenge in the clinical setting.

The next question we should be asking is whether we are being inappropriately optimistic about this field of research. Revolutionary advances catalyzed by the Human Genome Project decades ago and fueled by the more recent Precision Medicine Initiative have provided full molecular characterization of patients through multi-omic approaches. To translate this data-based knowledge into clinical care, we need to appreciate education and training for all urologists, emerging younger health care professionals in urology, and patients. Training the next generation of clinicians to take on the role of connectors in multidisciplinary teams is a *sine qua non*. Integration of molecular data into electronic health records also remains in developmental infancy. To apply biomarker panel testing as part of the standard of care in the clinical setting, clinicians should be aware of sufficient evidence, cost, and turnaround time. We should make an effort to understand the power and limitations of current testing and the treatment landscape in order to make the best-informed decisions. It should also be emphasized that when communicating to patients, we should be clear that there are limitations to molecular testing and it may not always be a realistic option.

We also encourage traditional laboratories to start collaborating or working with functional genomics by implementing more holistic techniques, like untargeted

metabolomics, lipidomics, and glycomics. Cultural changes committed towards a nonhierarchical structure in the clinic are also needed. To be consistently biased for teamed science, heavy investment from leaderships of major health care centers on collaborative efforts among molecular pathologists, geneticists, biostatisticians, bioinformaticists, epidemiologists, and translational scientists are needed. In terms of the clinical setting, several free websites currently offer frequently updated and deeply curated information on treatment variants that can be very useful in ascertaining whether a particular therapy is right for a patient.

The last, but not least, point of consideration is privacy and security policies in the context of precision medicine. Ethical and legal frameworks utilizing these datasets for discoveries should be discussed. The efforts, including governance, transparency, participant empowerment, respect for preferences, data sharing, access, use, data quality, and integrity, will provide assurance that individual data are protected and used in an appropriate and ethical manner. This is extremely important and we must continue to work

on these issues to sustain public trust and maximize the benefits of precision medicine in urology.

## CONFLICTS OF INTEREST

The author has nothing to disclose.

## ACKNOWLEDGMENTS

Dr. Jayoung Kim (Associate Professor of Surgery, Cedars-Sinai Medical Center, University of California Los Angeles, Los Angeles, CA, USA) is gratefully acknowledged for her contributions to article drafting and scientific discussion with this editorial.

Wun-Jae Kim

ORCID: <http://orcid.org/0000-0002-8060-8926>

Department of Urology, Chungbuk National University

College of Medicine, Cheongju, Korea

E-mail: [wjkim@chungbuk.ac.kr](mailto:wjkim@chungbuk.ac.kr)