Precision medicine and the transformation of healthcare delivery

Executive Summit 2017

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Summary

The ideal of treating patients in a targeted manner based on their individual condition (in other words, using the right treatment for the right patient at the right time) is becoming more possible than ever before. The rapidly increasing availability of genetic information and the growing "health data lake" – which includes clinical, imaging, and laboratory data, as well as information about the individual patient's lifestyle and everyday activities – provide a foundation to create a personalized risk profile to help make clinical decisions and more-precise recommendations for prevention. In the future, doctors will be able to increasingly align their behavior with patient-specific health data clouds, while the patients will be put in the position of taking control of their own health by, for example, self-tracking their lifestyle and acting as partners in the healthcare process. It is clear that data-based precision medicine like this requires a change in care delivery. (Semi-)automated IT solutions that are standardized, able to function in real time, fit for everyday use, and user-friendly will need to be developed to even use this growing knowledge and these capabilities to analyze individual differences practically – without overwhelming the treatment providers and doctors with an overflow of information. The structural integration of innovative technologies in clinical workflows is a prerequisite for precision medicine. A good example of this is the transformation of radiology into a quantitative discipline that is supported by statistical methods, interoperable databases, and artificial intelligence. This approach, which is called "radiomics" and which will also change the work and training of radiologists, could significantly contribute to individualizing the clinical management and choice of treatment in patients with various diseases.

One size does not fit all

It is an ideal of medicine to treat patients based on their individual conditions. One classic example is blood transfusions, in which patients receive different blood donations depending on their blood type. However, the possibilities for genuine precision medicine are now much greater than ever before. On the one hand, the availability of genetic information has grown exponentially in recent years, diagnostic and therapeutic procedures have been diversified, and molecularly designed medication and companion diagnostics have entered the realm of clinical practice. On the other hand, innovative IT approaches and artificial intelligence methods have become increasingly available to combine comprehensive databases, electronic patient records, and even lifestyle information to make personalized risk profiles and utilize them for clinical decisions.

Ever since former U.S. President Barack Obama announced a "Precision Medicine Initiative" in 2015, the idea of trying to use the right medical measures for each patient at the right time – and avoid unnecessary or even harmful measures – has been gaining ground in the public health sector. One size does not fit all.

It is clear that data-based precision medicine like this requires a change in healthcare delivery. (Semi-) automated IT solutions that are standardized, able to function in real time, fit for everyday use, and user-friendly will need to be developed to use this growing knowledge and these capabilities to analyze individual difference practically – without overwhelming the treatment providers and doctors with an overflow of information. The integration of innovative technologies in clinical workflows is a prerequisite for precision medicine. Personalized care opens the opportunity and need to involve patients more as partners in prevention and treatment.

Leveraging the data lake

One example of the range of precision medicine is the analysis of patient-specific genetic data. "We can start matching drugs with the genome," comments Amy Compton-Phillips, Chief Clinical Officer and Executive Vice President at Providence St. Joseph Health, a large not-for-profit health and social-services system in the U.S. Thanks to the rapid reduction in sequencing costs in recent years, targeted treatment decisions based on genetic analyses could soon become the common clinical standard.

However, genetic information is only one source of data. Large healthcare service providers like St. Joseph Health have already established a growing "data lake" that includes clinical and laboratory data, information about a patient's microbiome or lifestyle data, explains Compton-Phillips. In the future, it will be crucial to transform this data lake into a database of actionable possibilities and create personalized and dynamically changing data clouds for every patient.

"We are moving towards personalized plans of care – not only for treatment, but also for prevention," underscores Compton-Phillips. Using smartphone apps, patients can already receive health information that is tailored to their current situation, such as during the various stages of pregnancy or development of a newborn. In the future, integrated data analyses could also better integrate genetic factors and a patient's individual circumstances and provide continuously personalized healthcare counseling. Compton-Phillips claims that patients will increasingly be able to take control of their own health by self-tracking their lifestyle – which is completely in line with the transformation to predictive, preventative, personalized, and participatory medicine ("P4 Medicine").

Other experts also expect that personalized medicine will push the role of the patient as a partner to the foreground and make patients co-pilots instead of just passengers in the healthcare system – a model also known as "co-care." Innovative IT platforms that provide relevant data analyses in real time should prove to be effective tools in supporting flexible, customized, and virtually enhanced doctor-patient interactions and actually make personalized decisions possible.

Making better use of available knowledge

The importance of intelligent IT infrastructures can already be seen in the status quo of medicine. "We are still doing things to patients that we know they are not benefiting from," explains Ramin Khorasani, Professor of Radiology at Harvard Medical School and Distinguished Chair for Medical Informatics at Brigham and Women's Hospital in Boston, Massachusetts. One reason for this less-than-ideal healthcare is due in no small part to the fact that no doctor can consider the enormous amount of best available knowledge in their decisions. Best practice is not a universal standard. In fact, the variety of healthcare services and lack of standardization are common. It is not just crucial to integrate innovations and new discoveries into healthcare for better, more precise medical care, but also to apply what is already known. "Failure of execution on what we already know is a major challenge in transforming healthcare delivery today," says Khorasani.

However, he adds that the solution to the problem is not a rallying cry to doctors along the lines of "working harder and smarter." Instead, personalized, value-based care must be incorporated into clinical workflows with the help of intelligent IT solutions. It is important to recognize that IT is not just helpful per se and, for example, even electronic medical records can become a contributing factor of burnout among doctors due to an inefficient architecture, deflection, or the pressure of documentation. Digital platforms that make clinical work much easier and more convenient, rather than more difficult through additional information overflow, are therefore important. "The physician is a key stakeholder in healthcare who has needs himself," stresses Khorasani.

One instrument that might help is the Harvard Medical School Library of Evidence, which is a publicly available decision-making tool for radiological diagnostics that can be integrated into existing IT systems. The repository compiles guidelines from specialist associations, expert advice, and local best practices in a curated process and in an easy-to-use way. The doctor using the repository can check, for example after a mild head injury, under what circumstances a head CT is necessary and appropriate and when it is not, using concrete clinical scenarios in an app. The wealth of existing knowledge can therefore be directly applied to individual patients.

Towards precision diagnostics and quantitative imaging

Generally speaking, one core element of precision medicine is specific and individualized diagnostics, because targeted and effective treatments are usually only possible through a correspondingly precise diagnostic procedure and reliable risk prediction.

One example is an implantable cardioverter defibrillator (ICD) in patients with heart problems. Around 300,000 people die of sudden cardiac arrest every year in the U.S. alone, which in many cases can be prevented with an ICD, explains Lawrence Chu, Executive Director of Stanford Medicine X, an initiative that aims to explore how emerging technologies will shape future healthcare. Conversely, 90% of the patients who have an ICD do not benefit from it, which leads to the paradoxical situation that too many and too few of the devices are being used at the same time, states Chu. This dilemma can only be solved through the more precise selection of patients and those at risk of sudden cardiac arrest (for example, through better diagnostic stage algorithms with blood tests and imaging techniques as well as new digital analysis tools).

In contrast, precision diagnostics is already close at hand or already reality in other areas such as oncology. "For example, there are emerging approaches to monitor chemotherapy resistance in cancer patients through analyzing DNA from blood samples, and to tailor clinical decisions accordingly," explains Chu.

The transformation of radiology towards a quantitative discipline supported by statistical methods and artificial intelligence (AI) could prove to be even more far-reaching. The approach, which is called "radiomics," promises a whole new level of individualized diagnostics and therapy planning using extensive data analyses. "We are seeing a mathematical revolution in radiology," says Stefan Oswald Schönberg, Managing Director of the Institute for Clinical Radiology and Nuclear Medicine at University Hospital Mannheim, Germany. This transformation process will have a lasting impact on the work (and training) of radiologists in the years ahead.

For example, many tumor clusters are characterized by their high genetic heterogeneity with various tumor cell lines (clones), which often have varying degrees of aggressiveness, respond differently to treatments, and have compositions that can be decisive factors for the success of the treatment. The key here is to offer a precise image of the disease – through quantitative analyses of imaging information as well as AI-based, integrated interpretations of clinical, genetic, and imaging data – and then to tailor the treatment more precisely to the individual patient, explains Schönberg.

According to Schönberg, the quantitative data from high-resolution CT images can be used for therapy planning for even chronic illnesses such as COPD (chronic obstructive pulmonary disease), especially if the data correlate with information from other procedures, such as a pulmonary function test (body plethysmography) and are evaluated together. Overall, this further developed radiology in the era of precision is expected to play a leading role in making the best medical decisions for individual patients.

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Published by Siemens Healthcare GmbH · Printed in Germany · 5719 0118 online ·