The paradigm of “personalized medicine” has gained broad popularity in the past decade due to the fact that convincing evidence was brought that the concept “one fits all” should be overwhelmed by the so-called patient-tailored approach in health care [1, 2]. Although the term personalized medicine is repeated like a mantra in scientific papers, meetings, media, and even the Internet, its exact definition is vague and remains almost unclear for many. A recurrent but rather incomplete definition is “prevention, diagnosis, and therapy of a certain disease according to an individual genetic profile” [3]. Indeed, the complete picture cannot be limited to genomic analysis, wherein molecular methods that make personalized medicine possible include testing a wide spectrum of biological pathways, thus embracing gene polymorphisms and expression, epigenetics and metabolomics, along with identification of therapies aimed to target molecular mechanisms that are responsible for several human disorders, including cancer [4], cardiovascular disease [5], and systemic infections [6], among others. The current designation proposed by the EU Commission seems more appropriate, wherein personalized medicine is defined as a medical model using molecular profiling for tailoring the right therapeutic strategy for the right person at the right time, and/or to determine the predisposition to disease and/or to deliver timely and targeted prevention. Throughout this ample picture, laboratory diagnostics is seemingly playing a prominent – even predominant – role, which is probably much larger than that of any other branch of science and medicine.

Despite the fact that transition from traditional medicine toward personalized health care seems now a virtually unavoidable journey, some important challenges emerge. The leading principle is indeed represented by the convincing awareness of the pivotal role that laboratory professionals should play in translating theory to practice. To assess whether laboratory medicine would be able to help develop innovative diagnostic tools, skill, and guidance about personalized medicine, the joint working group “Personalized Laboratory Medicine” of the EFLM (European Federation of Clinical Chemistry and Laboratory Medicine) and ESPT (European Society of Pharmacogenomics and Personalised Therapy) has developed and disseminated to 48 laboratories from 18 European countries an interesting questionnaire entitled “Is Laboratory Medicine ready for the era of Personalized Medicine?”, the results of which are published in this issue of Clinical Chemistry and Laboratory Medicine [7]. Encouraging information emerged from the results of this survey, indicating that laboratory professionals not only do acknowledge that personalized medicine may be regarded as an innovative and promising health model, but also that laboratory diagnostics will play an essential role for supporting a widespread implementation of this new approach to health care in the clinical setting. Now, despite the fact that we can probably consider these aspects for granted, it seems necessary to move further and put forward another crucial question, that is, “how should we reach the goal”? There is no single right track, indeed, wherein moving from simple theory to daily practice entails a challenging and multifaceted enterprise, in which many aspects should be considered (Figure 1). First and foremost, the gap between basic research and clinical practice should be narrowed, with identification and validation of biomarkers (or panels of biomarkers) that may realistically generate medical knowledge and be effective to impact on the outcomes (i.e., disability, morbidity, or mortality) [8]. A number of well-designed and large international initiatives are under way, and results are expected to be delivered soon [9]. A second necessary step entails the development of robust and reliable assays that may be able to precisely identify biological patterns associated to (or responsible for) the most prevalent human disorders [10]. Once these basic requirements are fulfilled, we should then overcome the traditional theory of reference ranges, toward a new concept of individual reference values that would allow a more reliable risk prediction in the single patient [11]. Then, the awareness of the increasing importance of personalized medicine should not be regarded as an ending point but as just a beginning, wherein its use should be much broadened
in clinical practice, over the strict boundaries of basic research where it is currently confined [12]. A last but essential aspect in a world with limited resources is the appropriate use of innovative (and often expensive) tools such as genomic, transcriptomics, pharmacogenomics, proteomics, and metabolomics, in order to prevent that precious human and economic resources are dissipated for targeting theory rather than patient-centered practice [13]. As we are entering the era of the so-called direct-to-consumer marketing of personalized medicine [14], in particular the offer over the Internet of direct-to-consumer genetic testing services [15], further considerations should be made. Test relevance and accuracy are still fundamental issues, as emphasized by the decision of the US Food and Drug Administration to ask a direct-to-consumer genetic testing company to stop selling its personal genetic testing kits to consumers owing to significant concerns regarding clinical basis, relevance, and accuracy of results [16]. Therefore, the need for training of physicians and health-care professionals to correctly request and interpret genomic data and to integrate genomic information with the patient’s clinical condition and all other variables in an expert and systematic way should be regarded as additional barriers to broader adoption of personalized medicine [15]. Nevertheless, a simple approach can be applied, entailing the use of the right test, with the right method, at the right time, to the right patient, at the right cost, and for the right outcome [17].

References


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