



Editorial

Molecular diagnostics and personalised medicine

Frank Desiere, frank.desiere@roche.com and Vincenzo Romano Spica, vincenzo.romanospica@uniroma4.it

This special issue of New Biotechnology is focused on molecular diagnostics and personalised medicine and appears at an epochal moment in the development of the field. The practice of medicine is taking a significant and irrevocable turn towards personalisation, due to the great progress in areas such as genomics, pharmacogenomics and molecular diagnosis. It becomes increasingly apparent that to deliver the promise of personalised treatments, more and more novel medicines discovered today will be presented together with innovative companion diagnostics.

The contributions to this volume touch on many disciplines, ranging from cell biology to genetics, immunology, molecular diagnostics, pharmaceuticals and economic issues. The contributions of clinicians and basic scientists are synergistically presented to underline better the wide spectrum of studies that can contribute to the new field of personalised medicine. The promising perspectives of individualised treatments are related not only to higher effectiveness, but also to increased efficiency. This is relevant not only for the individual patient, but even more so for the general public, within a wider economical perspective where resources are limited and it becomes more and more mandatory to close the gap between social costs and benefits. This approach follows the steps of a stratified and individualised medicine and finds its final goal in an individualised healthcare.

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It is true that over the last decade, personalised medicine has fallen short of the predictive and preventative healthcare paradigm it once promised, as discussed in a recent Nature Biotechnology editorial [1]. In our opinion this was primarily due to premature high expectations of the field and asynchronous progress in relevant areas.

We would like to highlight three major drivers of change that are among others that will take the field forward.

Economics

2012 marks a new and different period for the pharmaceutical industry as the previous drug business model seems to be set to change. Most evident is the industry transformation by the profound organisational changes in many of the leading companies over the past few years. In addition, several pharmaceutical companies are also increasingly targeting smaller indications for which patient numbers may be smaller, stimulated by the Orphan Drug Act [2]. This trend, among others, promises to improve the lives of many patients that have previously not been treated with the best-suited medicines, or for whom there were no effective treatment options at all [3]. Therefore, this new model will drive innovation.

Healthcare spending in relation to gross domestic product (GDP) is becoming increasingly burdensome to country economies which are already financially strained. Together with the ageing population and increasingly complex yet useful medical interventions, there are increasing constraints on the economics of the industry. There is evidence today that competition in the industry is rising with a growing number of players competing for constrained resources [4].

However, in this environment, two pharmaceutical business models will certainly continue to thrive. There is a place in the future for companies that provide low cost solutions (generics), as they provide the most cost efficient solutions for essential off-patent medicines. In addition, those firms that will be able to generate highly differentiated and value-adding products through personalised medicine will continue to be successful, as they commercialise products supported by strong clinical and clear economic data that can truly justify higher costs when payers start pushing back on reimbursement. As a result, the personalised medicine approach will become mainstream for most new, highly differentiated innovative drug developments.

Regulation

The acceptance levels, of both the regulatory authorities and of patients, for drug side effects have been going down over the last years. This development has led to the development of drugs that are safer and better suited to a particular group of patients, with fewer side effects and well-demonstrated efficacy.

Because of this concern, among other reasons, the number of new drug approvals decreased in the last decade, while their review time increased sharply. 2011, however, was marked by a notable development. Two new drugs [5,6] were approved well ahead of their projected review timelines. These drugs were co-developed alongside their companion diagnostic tests. The resulting clinical data were sufficiently convincing for the FDA to fast-track the approvals.

By contrast, also last year, the FDA rejected a new drug for treatment of a subset of leukaemia patients whose tumours had a particular genetic mutation [7]. The main problem was not the drug itself, but rather that the company had not specified a companion test that could reliably detect the mutation so that the drug could be given to the patients it is intended to help. In such circumstances, companion tests are becoming almost gatekeepers to the drug.

Currently, it is often not enough for pharmaceutical companies merely to bring a drug to market. Regulators and insurers are also requesting the companies to develop tests to pinpoint which patients are most likely to benefit, thereby sparing others from needless side effects and expense. Personalised treatments may thus become essential in the future.

Innovation

Molecular technologies such as anatomic pathology and sequencing technologies are becoming commonplace in medical research, and some have already made their way into clinical practice. DNA sequencing is in fact becoming so affordable and accurate that it can now be envisaged to use this technology more broadly in medical research than ever was possible before. We can speculate that the technology will also be more prominently used as an *in vitro* diagnostic in the future.

As a result of these ground-breaking changes, we have brought together key opinion leaders to contribute to this special issue and discuss the many aspects and complexities of personalised medicine in a cross-disciplinary manner. This broad contribution underlines the fact that the future of personalised medicine will not depend just on a few scientific breakthroughs. The success of its promise will rely on coordinated progress in all related disciplines to capture, convince, reward and motivate all parties involved in molecular diagnostics, drug development and drug delivery, as well as in the regulatory issues of medicine, to move forward in a concerted way.

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The Forward of this special issue, kindly provided by Dr. Leroy Hood, adds clarity and vision for the challenging way ahead. A main conclusion emphasises the importance of harmonisation between scientific achievements, clinical practice and healthcare systems. This approach is considered essential for an effective application and progress of personalised medicine. Indeed, the availability of new tools for therapy individualisation is based not only on the availability of laboratory biomarkers, but also on appropriate data systems, capable of managing the complex network of interacting factors.

In conclusion, the guest editors of this special issue are confident that this collection of highly relevant and impactful contributions will cater to many diverse audiences, be they in natural sciences, medicine, public policy or other fields with relevance to personalised medicine. We are grateful to have achieved such a high quality collection of papers and would like to thank the authors for their contributions. We would like to encourage the whole field to continue the progress in personalised medicine, and to further realise its promise for the improvement of patient lives.

References

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Frank Desiere

Roche Diagnostics, Basel, Switzerland

Vincenzo Romano Spica

Unit of Public Health, University of Rome "Foro Italico", Rome, Italy