

Reaching a Personalized Medicine Era: The Dream of the Drug Market

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Abstract

Personalized medicine, the study of the influence of a patient's genetic makeup on their disease susceptibility, prognosis, or treatment response (efficacy and safety), is actually in the spotlight. This field is expected to allow us to have effective and safe medication to targeted patients with appropriate genotypes.

Keywords: Personalized Medicine, Pharmacogenetics, Clinical Outcomes, Human Genomics, Drug Response

In the last few decades, the practice of medicine has seen swift changes, as well as its visualisation in the near future. It was designed and focused on serving the community and helping people in need. However, it is not a secret that there is a huge business around this labour and the economic interest of a diverse industry in the field.^{1,2}

Not intending to generalise, many have observed in daily practice a comparable trend with modern society. A phenomenon including both patients and health personnel, where there is a demand for health services, a growing supply, and a considerable revenue. Basic market economics, right?³

Not that simple.

It would be the triumph of basic sciences to explain each disease under a biological substrate, minimising the involvement of other factors. A definitive targeting of biological research would be the key to unlocking knowledge. What is certain is that this approach has transformed pharmacotherapy, treatment alternatives and prognosis.^{2,3,4}

Early physicians had little to nil information on what today we call aetiology, pathophysiology and therefore treatment. Patients were rarely relieved due to human intervention. Trepanations were frequently performed in the Classical and Renaissance periods and although having modern indications (decompressive craniotomy), its uses and technique were at best questionable. Belief and verbally transmitted understanding of a handful of medicinal plants whose effect were known empirically were standards of care.⁵

These times have changed, the pharmaceutical industry is a pillar of the economies in many countries, and the number of transactions and cash flow that they move are beyond the wildest dreams of the first physicians. Born each year, thousands

of new pharmaceutical companies develop and market new drugs and medical supplies.^{1,6}

As advocated by experts, pharmaceutical and medical supply companies are considered one of the safest businesses nowadays, with everyone being a potential consumer/patient. It is the race for continuous development of new drugs to its current rate that guarantees soon we will have more drugs and procedures available. The drug industry may be easily overloaded by an oversupply of organic compounds and procedures to patients.^{2,4,6}

This pharmaceutical industry thriving is widening its horizon. Personalised medicine, the study of the influence of a patient's genetic makeup on their disease susceptibility, prognosis, or treatment response (efficacy and safety), is actually in the spotlight. This can be assessed in different ways, being preventive and/or therapeutic.⁷

In the preventive field, preconception screening studies have been unravelling genetic disorders, as recommended by different guidelines such as those of the American College of Medical Genetics, which are designed for individuals with known genetic conditions or high-risk patients who wish to become pregnant.⁸

In the therapeutic field, pharmacogenomics can aid in the identification of alterations of Single Nucleotide Polymorphism (SNPs) that affect the function or expression of proteins associated with pharmacokinetics or pharmacodynamics of different drugs. In recent years the research community has doubled efforts in personalising certain therapies. Hormonal therapy in breast cancer has been from the beginning a receptor-guided therapy, especially with ER (Oestrogen Receptor) therapy. Initial clinical results of trials conducted so

far have allowed to establish single therapies regimens with Tamoxifen or combined with Arimidex.⁹

Another model of the advances in this arena is reflected in the new alternatives for prostate cancer. This hormone-dependent tumour has demonstrated recurrent alterations in the androgen receptor and its pathway. In specific patients the disease can be found in Castration-Resistant Prostate Cancer (CRPC), a lethal clinical state in which the tumour has developed resistance to androgen deprivation therapy. This clinical scenario is commonly established in advanced or metastatic prostate cancer patients. The genomic landscape of localised prostate cancer has been well defined, describing putative pathogenic BRCA2 germ line mutations as well as somatic and germ line DNA repair alterations found such as BRCA1, CDK12, FANCA, and RAD51B. Furthermore, the research advances described above can allow clinicians to determine treatment, therefore achieving better outcomes.¹⁰

It is unquestionable that personalising treatment will improve clinical outcomes for patients in the near future and help achieve a more effective use of available health care resources. The next challenge for scientists and researchers is to demonstrate with strong evidence the clinical and cost-effectiveness to support the use of personalised medicine and its implementation in different health care systems around the world.^{2, 3, 5}

In conclusion, individual patient variability currently studied in drug efficacy and drug safety has represented a major objective in current clinical practices. Years of research results have converged in progresses in pharmacogenetics and human genomics that have dramatically accelerated the discovery of genetic variations that potentially determine variability in drug response, providing better clinical outcomes for patients. The future in this field is expected to allow us to have effective and safe medications to targeted patients with appropriate genotypes.

Competing Interests

None declared

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