

# Towards precision medicine

Many decades ago, physicians came to appreciate the uniqueness of every patient and the need for specific personal treatment and care. It is largely because every person has a unique variation of the human genome. Most of these variations compose personal individuality and have no effect on health. However, individual factors of behavioural and environmental nature might have a harmful effect on the genome, thereby causing health problems. State-of-the-art molecular and genetic techniques enable the evaluation of individual susceptibility to various risk factors, timely identification of inherited disease-causing mutations, a more accurate diagnosis of sporadic diseases, and the development of more efficient drugs and therapeutic strategies.

The phrase “personalized medicine” appeared at the beginning of this century and has since then been defined in various ways. In 2005, the US Food and Drug Administration (FDA) defined personalized medicine as “the best medical outcomes by choosing treatments that work well with a person’s genomic profile, or with certain characteristics in the person’s blood proteins, or cell surface proteins”. Later, the FDA issued recommendations regarding the integration of genetic and biomarker information for clinical use and drug development. From 2015 onwards, after the White House press release, the term “precision medicine” has replaced “personalized medicine”. As defined in the press release, the major goal of precision medicine is to enable clinicians to provide “the right treatment at the right time to the right person”, taking into account the health history, genes, environments, and lifestyles of individuals. By this definition of precision medicine, one means that the best health care solutions are offered for well-defined and carefully selected groups of patients and not simply to one particular person. With the help of genomics, proteomics, or panels of selected biomarkers, individuals are classified into subgroups that differ in their susceptibility to a particular disease and in their response to treatment.

Precision medicine integrates various research disciplines, including molecular biology, immunology, nanotechnologies, genomics, and epigenomics. In tandem with clinical practice, it strives to advance medicine and patient care. The present issue of AML provides several publications that nicely illustrate our progress toward the implementation of precision medicine in Lithuania. The paper by Gasperskaja and Kučinskis reviews the main technologies and tools used for functional genome analysis, while Lachej et al. emphasize the importance of knowledge of molecular pathways causing gynaecological malignancies. The methods of precision medicine could assist in the assessment of inherited or acquired risk factors for a number of illnesses, including immune dysregulation syndrome hemophagocytic lymphohistiocytosis (Bareikienė, Rascon) or inherited cases of testicular cancer (Ulyté et al). Timely identification of inherited disease cases can tailor individual preventive treatment and save many lives.

The development of novel diagnostic methods, the usage of target-directed treatment means, and the implementation of harm-reducing laparoscopic surgeries are the key attributes of modern medicine (Dulskas et al.).

Research in precision medicine has already led to new discoveries: the invention of new diagnostic tools and the development of novel genetic knowledge-driven treatment strategies, among others. Precision medicine offers the safest and most effective treatment for a specific disease and suggests tools for non-invasive follow-up based on molecular testing of disease outcomes. Moreover, precision medicine

can greatly support the advances of preventive care. For instance, many women with familial history of breast or ovarian cancer are genotyped for mutations in the *BRCA1* and *BRCA2* genes. Early genetic profiling of the risk can assist in timely prescription of preventive means and prolong the healthy lifespan. During this era in medicine, screening programmes for early detection of cervical, breast, colon, prostate cancer, and cardiovascular diseases are proposed and launched that employ modern visualization and molecular diagnostic tools to reduce the burden of the most lethal illness of our days. New techniques and biomarkers are coming into clinical practice to aid early detection and specific treatment of many other lethal and disabling diseases, including skin, stomach and lung cancer, diabetes, arthritis, etc.

However, this period of active transfer of technology and knowledge into the clinics calls for some new policies. There is an urgent need for continuous training of clinicians, for recognition of new interdisciplinary professions, and for introduction of novel university courses. One would expect the latter to train next-generation physicians to be able to interpret and use the broad clinical, immunological, genetic, metabolic, and other personal data of each patient to tailor proper medication, therapy, or preventive means.

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