

# Preface

Traditionally, medical research comprised of the identification of the pathological causes of a disease, its epidemiology and empirical investigation of treatment response. Intensive genetic research, marked by the completion of the human genome project in 2003, heralded a new era in medical research. While epidemiology and gross pathology are still mainstay useful tools, genetics and genomics have gradually been shown to increase the resolution of drug response research, showing great potential in also informing and identifying the role of genes and their encoded products in the pathophysiology of diseases. This information is already being applied to effective early diagnosis, better risk assessment (prognosis), as well as targeted effective and safe treatment allocation (prediction and monitoring).

Genetic testing and genomics support personalised medicine by translating genome-based knowledge into clinical practice, offering enhanced benefit for patients and health-care systems at large. Current routine practice for diagnosing and treating patients is conducted by correlating parameters such as age, gender and weight with risks and expected treatment outcomes. In the new era of personalised medicine the healthcare provider is equipped with improved ability to prevent, diagnose, treat and predict outcomes on the basis of complex information sources, including genetic and genomic data. The support of regulatory bodies and policy makers internationally has been critical for the rapid translation of personalised medicines into the clinic. Notwithstanding, inequality in the utilisation of targeted therapies in different health care systems across the world exists, and ethical considerations, as well as economic cost-effectiveness analyses are in need to inform decision making. In addition to the benefits of pharmacogenomics in diagnosis and treatment, prevention of illness using genomic information is important to reduce the burden on the healthcare system, a methodology proven effective in many therapeutic areas, but, paradoxically still facing challenges in others. In current settings screening programmes (e.g. BRCA1/2 screening) address this by identifying susceptible families and preventive measures or ensuring appropriate treatment at the earliest stages of disease, hence increasing health management effectiveness.

The integration of pharmacogenomics into the various health care systems have been the responsibility of the respective national health authorities, which in turn follow recommendations by leading regulatory bodies such as the European

Medicines Agency and the US FDA. The dynamics and logistics of this integration therefore vary substantially across the globe. To this end, implementation of pharmacogenomics is an important component of PPPM (Predictive, Preventive and Personalised Medicine), which is the main focus of the European Association for Predictive, Preventive and Personalised Medicine (EPMA). Since 2009, EPMA (<http://www.epmanet.eu/>) embarked on various initiatives to promote PPPM including the launch of the EPMA Journal to ensure dissemination of current aspects of PPPM, and the organisation of the first World Congress in September 2011 bringing together participants from over 40 countries to discuss education, policy and implementation of PPPM. In addition, EPMA took the initiative to publish a series of books in advances in PPPM, including the present one entitled “**Preventive and Predictive Genetics: Towards Personalised Medicine**”. The editors take this opportunity to thank all contributing authors and trust that the content meets the expectation of all readers.

The Editors

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