#### Published online 2013 November 20.

Editorial

## Personalized Cancer Medicine: a Future Direction of Personal Genomics

# Ehsan Ghayoor Karimiani<sup>1,\*</sup>

<sup>1</sup>Department of Medical Genetics, Mashhad University of Medical Sciences, Mashhad, IR Iran *\*Corresponding author:* Ehsan Ghayoor Karimiani, Department of Medical Genetics, Mashhad University of Medical Sciences, Mashhad, IR Iran. Tel: +98-9153102327, Fax: +98-5118002287, E-mail: ghayoure@mums.ac.ir.

Received: September 26, 2013; Accepted: October 9, 2013

Personalized medicine is significantly changing the fundamentals of healthcare. Genetic diagnostic tests are used in various parts of medicine from initial screening to the diagnosis and finally treatment. Investigations in fields of drug development indicate that oncology may lead other therapeutic areas in the number of targeted therapies on the marketplace as well as in the pipeline, expecting that within a few years, all oncology medications will have a related diagnosis. It has been shown that personal genomics will influence other important therapeutic areas, including the clinical decision making. In conclusion, personalized medicine is just getting started for hematology and respiratory medical therapies, as well as oncology.

Keywords: Individualized Medicine; Genomics; Molecular Diagnostic Techniques

### 1. Personalized Medicine and Molecular Genetic Diagnostics

Personalized medicine (PM) is considered as a medical model that strongly suggests the customization of healthcare, based on the clinical judgments, and/or medications being tailored to individual cases. Genetic information has played a pivotal role in the convinced aspects of personalized medicine, and nevertheless the term was initially applied in the context of medical genetics (Although it was broadened to include all types of personalization measures) (1). Medicine has always been characteristically "personal" to every patient, however; PM commonly signifies the use of some types of innovative enabling technology at a level of personalization not previously feasible or achievable. Personalized medicine, also referred as genomic medicine, is significantly changing the fundamentals of healthcare. By coupling the strength of genetic testing, doctors may make more informative evidence-based medical decisions to improve target treatments and drug therapies. This will obviously result in a better healthcare outcome (2).

Molecular genetics diagnostic tests are used in various parts of medicine – from initial screening to the diagnosis and finally treatment. Researches in those fields of drug development indicate that oncology may lead other therapeutic areas in the number of targeted therapies on the marketplace as well as in the pipeline, expecting that within a few years, all oncology medications will have a related diagnosis. Other important therapeutic areas in which personalized medicine will influence on clinical decision-making include cardiovascular, immunologic and neurologic therapies (3). However, personalized medicine is just getting started for hematology and respiratory therapies, as well as virology.

It is now well-understood that personalized medicine is becoming an integral part of the healthcare, changing the patient care landscape. There are several groups such as DNA Direct Genomic Medicine Network, which consists of different specialized hospitals, offering both physicians and patients to embrace the power of personalized medicine, to make more validated medical decisions and to improve the treatment. Nevertheless, it helps physicians and clinical staff to navigate the complications of genetic testing through access to the facilities needed to take the "right test, at the right time, for the right patient".

Important cellular functions including growth, apoptosis, biological movement and protein co-localization plus differentiation, etc. significantly modulate the process defined as signal transduction. This process is almost completely epigenetic and ruled by the protein enzyme activity. Based on genomic mutations, diseases such as cancer are functionally marked as dysfunctional protein signal transduction. Pharmaceutical interventions, therefore aim to control the aberrant protein activity, not the genetic abnormality. Hence, the comparative analysis of gene and protein expression, called genomics and pro-

Implication for health policy/practice/research/medical education:

Personalized medicine, referred as genomic medicine, is significantly changing the fundamentals of healthcare. By coupling the strength of genetic testing, doctors may make more informative evidence-based medical decisions to improve target treatments and drug therapies. This will obviously result in a better healthcare outcome.

Copyright © 2013, Razavi Hospital; Published by Kowsar Corp. This is an open-access article distributed under the terms of the Creative Commons Attribution License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original work is properly cited.

teomics have shown little concordance between the two 'omics'; as a result, some scientists now believe that the direct analysis of proteome may be required (4).

### 2. Molecular Cancer Medicine in Healthcare

Cancer genetics is a particular field of genomics that is focused on the risks for hereditary cancers. Currently, there are very few predisposition syndromes in which an allele separates in an autosomal dominant fashion, to increase the risk for specific cancers. It is well estimated that familial cancer consists of approximately 5 - 10% of all cancers. However, other genetic variants influencing delicately on the cancer risk, may be used more precisely in cancer risk assessment in patients without a strong family history. Examples of personalized cancer management include testing for disease-causing mutations in the BRCA1 and BRCA2 genes, which are strongly associated with hereditary breast-ovarian cancer syndromes. Detection of such a disease-causing mutation can highlight the "at-risk" cases in a family and may lead to prompt individualized prophylactic treatment including elective mastectomy and removal of the ovaries. This diagnostic test may be involved in complicated personal decisions in the context of thorough genetic counseling sessions (5). Therefore, more comprehensive molecular classifications of breast tumors may lead to the future tailored treatments. Targeted therapy are scientifically designed to target abnormal molecular pathways in a subgroup of patients with a given cancer diagnosis (6). For example, trastuzumab (marketed as Herceptin) is currently used in the treatment of patients with breast cancer in which HER2 protein is amplified.

Minimal Residual Disease (MRD) diagnostics are also used to quantify and monitor residual cancer, enabling detection of tumor markers before the clinical relapse. This has extremely helped physicians in making clinical decisions sooner than previously possible. Tyrosine kinase inhibitors such as Imatinib (marketed as Gleevec®) or Dasatinib (Sprycel®) have been developed to treat chronic myeloid leukemia (CML), in which the BCR-ABL fusion gene (the product of a reciprocal translocation between chromosome 9 and chromosome 22) is present in > 95% of cases. It has been shown that, in these cases, there is a hyperactivated abl-driven protein signaling. These medications are specifically inhibitor of the Abelson Tyrosine kinase (ABL) protein and are therefore typical example of "rational drug design" based on the science and knowledge of the disease pathology (7).

#### 3. Concluding Remarks

In general, personalized medicine and its related mo-

lecular genetic services are integral components of enlightened, forward-thinking institutions. The aforementioned services will undoubtedly provide hospitals with an opportunity to drive the downstream revenue. As medical scientist and geneticists in particular, have basically focused on innovation and personalized medicine, their leaders endeavor in genomics, proteomics and recently in metabolomics. With the advent of all new technology and diagnostic platforms available to the healthcare professionals, it will be critical to stay abreast of the new improvements. Low-cost Whole Genome Sequencing (WGS) is a significant goal as well, adding insightful new dimension to the personalized medicine in near future. Commercial kits and both providers and consumers in the healthcare will be challenged with how best to interpret the huge informative data extracted (8). As development in personalized medicine continues, patients benefit from the profound knowledge that 'omics' bring to medical care decisions and outcomes.

#### Acknowledgements

There are no acknowledgements.

#### **Financial Disclosure**

There is no financial interest to declare.

#### **Funding Support**

There is no funding or support.

#### References

- Schilsky RL. Personalized medicine in oncology: the future is now. Nat Rev Drug Discov. 2010;9(5):363–6.
- Karimiani EG, Day P. Personalised treatment of haematological malignancies through systems medicine based on single molecules in single cells. *Integr Biol (Camb)*. 2013;5(5):759–67.
- Esmaeili B, Rezaee SA, Layegh P, Tavakkol Afshari J, Dye P, Ghayoor Karimiani E, et al. Expression of IL-17 and COX2 gene in peripheral blood leukocytes of vitiligo patients. *Iran J Allergy Asthma Immunol.* 2011;10(2):81–9.
- Rosenblum D, Peer D. Omics-based nanomedicine: The future of personalized oncology. Cancer Lett. 2013.
- Natrajan R, Mackay A, Lambros MB, Weigelt B, Wilkerson PM, Manie E, et al. A whole-genome massively parallel sequencing analysis of BRCA1 mutant oestrogen receptor-negative and -positive breast cancers. J Pathol. 2012;227(1):29–41.
- Sandison HE, Usher S, Karimiani EG, Ashton G, Menasce LP, Radford JA, et al. PLK1 and YY1 interaction in follicular lymphoma is associated with unfavourable outcome. *J Clin Pathol.* 2013;66(9):764–7.
- Verma M, Karimiani EG, Byers RJ, Rehman S, Westerhoff HV, Day PJ. Mathematical modelling of miRNA mediated BCR.ABL protein regulation in chronic myeloid leukaemia vis-a-vis therapeutic strategies. *Integr Biol (Camb)*. 2013;5(3):543–54.
- Leary RJ, Sausen M, Diaz LA, Jr, Velculescu VE. Cancer detection using whole-genome sequencing of cell free DNA. Oncotarget. 2013;4(8):1119–20.