Review

Personalized medicine – the promised land: are we there yet?

The delivery of personalized genomic medicine (refer Table 1 for a comparison of genomic vs genetic medicine and box 1 for glossary) hinges on obtaining personal genomic data through genome-wide association studies (GWAS) or whole-genome sequencing. After the completion of the human genome project (see box 2 for human genome projects and its derivative projects) in 2003, there appeared to be a period of euphoric optimism that as soon as the cost of sequencing the whole human genome could be brought down to an affordable range, the promise of personalized medicine would become a reality. However, inasmuch as the miraculous technological advancements are making whole-genome data acquisition an inexpensive reality, we are also starting to appreciate that making sense of the enormous amount of genomic data is a far bigger hurdle. Issues, both scientific and ethico-legal, will have to be addressed as genomic data are been pushed for clinical and direct-to-consumer utilization.

Conflict of interest

Nothing to declare.

Scientific issues related to genomic data and their utilization

The usefulness of any clinical test or intervention is measured by its (i) analytic validity – that the results are accurate and reliable, (ii) clinical validity – that the reliable results are of consistent clinical significance and (iii) clinical utility – that the clinical significance is such that there is clear benefit for its utilization in healthcare in a cost-effective manner.

For genome-wide association studies (GWAS), the first and frequently cited success story is the discovery of strong associations of several genomic variants with age-related macular degeneration (AMD) (1–3). By virtue of these variants’ proximity to the gene encoding complement factor H (CFH), the role of CFH, a previously unsuspected player in this condition, was uncovered, and by inference, the complement network. In addition, deletions near CFH and its receptors CFHR1 and CHFR3 have been associated with reduced risk for AMD (3).

However, for most other GWAS, genomic variants noted are polymorphisms that maybe useful in haplotype identification in the study of genealogy or evolution, but their associations with a disease condition inconclusive and their functional significance elusive. Moreover, results from one GWAS are not readily replicated by independent studies (4–5). There have been efforts to integrate and standardize GWAS. But even when a GWAS study can be replicated, the associations are typically weak. Well before GWAS became a familiar term in research and clinical communities, clinical geneticists have faced the issues of low-penetrant mutations associated with monogenic Mendelian disorders. Using hereditary hemochromatosis (HHC) as an example, two mutations account for the vast majority of HHC cases (6); however, only a minority of people with...
disease-causing mutations will go on to develop the disease. Given the poor predictive value of a positive genetic test on the outcome, expert panels have largely discouraged genetic testing of asymptomatic healthy individuals, even those of high risk ethnicity, i.e. Northern European descent, where carrier frequency is as high as over 10% (7, 8). Instead, serum iron and ferritin levels and transferring saturation are better indicators of disease diagnosis and progression. A similar case can be made of type I Gaucher disease in the Ashkenazi Jewish population (8).

If genetic testing of low-penetrant mutations known to be a causative of certain diseases is discouraged, why would we want to test for genomic variants of even more modest association with a disease condition? Some argue it is because GWAS target common diseases. Cardiovascular diseases are the leading cause of death in developed countries, so even a weak association could have a significant population impact. Proponents (9) quoted studies on 12 genomic variants that were found to confer 1.6 times greater risk of heart attack in 10% of people of European origin over the population risk of same ethnicity, independent of other known risk factors. As the lifetime risk for heart attacks in this population is 49% for men over the age of 40, these 10% will have a lifetime risk of ~78%. However, as the increased risk is independent of known risk factors, there is no known intervention. Furthermore, one could use empiric risks for many common diseases simply based on family history without costly genomic testing. For example, high low-density lipid levels raise the risk of heart attack 1.3 times and having a first degree relative with type 2 diabetes or coronary artery disease increases one’s odds for either by 2.3–2.8 or 3.8, respectively (10). Indeed simulation studies have shown that the predictive value of genomic profiling may attain the same level as that of traditional risk in predicting cardiovascular diseases but not higher or high enough for predictive diagnosis (10).

Moreover, GWAS results will need to be validated by prospective studies (Fig. 1), which typically take years, if not decades.

Furthermore, common diseases being common, all individuals are likely to carry one or more risk alleles, and perhaps one or more protective or counter-risk alleles as well. If a million people were followed in a prospective study for 10 variants of four genome types each in a diploid

Fig. 1. Genome-wide association studies and the need for validation of them by prospective studies. DM2: type 2 diabetes mellitus; SNP: single nucleotide polymorphism.
genome, the number of potential combinations would be greater than a million. In other words, each individual likely would have a very unique combination profile (similar to DNA fingerprinting), thus making the comparison difficult, if at all possible.

Even when GWAS demonstrated a strong association, there may not be any clinical utility. Case in point is the finding that single nucleotide polymorphisms (SNPs) near the IL28B gene correlate strongly with responsiveness to interferon-α treatment for hepatitis C virus infection and 80% of individuals who were homozygous for these SNPs could be cured with the treatment. However the effect was not absolute, i.e. one cannot base treatment decisions on such genome info, and equally importantly there is no effective alternative treatment for hepatitis C virus infection (11–14). As a consequence, this genotype information has no bearing on treatment strategy so far.

What about whole-genome sequencing?

The haploid human genome contains $3 \times 10^9$ bases of DNA, of these, $\sim 1.5\%$ are genes that encode proteins, while another $\sim 3.5\%$ are non-coding sequences performing functions that we understand to some extent. They include gene-regulatory elements and chromosomal architectural and functional elements. Together they make up $\sim 5\%$ of the genome (15). The esteemed Encyclopaedia Britannica contains $\sim 4 \times 10^7$ words, all of which we understand provided we had the time to go through them. Now consider the fact that for the remaining 95% of the genome, or $2.85 \times 10^9$ bases, we do not yet know how to read their meaning.

A living organism is more than a genome. A human being develops from a single fertilized egg into a trillion-cell individual. Cells of identical genomes divide at different rate and differentiate into different cell types, skin, neuron, cortex or medullary tissues of a lymphnode, etc. They function differently and they are differentially sensitive to teratogenic effects of environmental toxins such as alcohol or anticonvulsants, at different developmental stages during embryogenesis, childhood and adulthood. They also show differential sensitivity to a genomic change throughout an individual’s life span. For example, a mutation in the RET gene renders very high risk for medullary thyroid cancer, but not retinoblastoma, whereas a mutation in the RB1 gene is associated with high risk of retinoblastoma but not medullary thyroid cancer even though both thyroid cells and the retinal cells have the same genome (16–18). Different cell types within an individual also have different life spans and different capacities to regenerate – skin cells regenerate all the time whereas neurons in our brain are as old as we are and hardly regenerate at all. How identical genomes lead to different cell types that function so differently? The overwhelming evidence of fetal alcohol syndrome makes it a legitimate assumption that two foetuses of identical genomes could develop quite differently with or without prenatal exposures to alcohol. In other words, environment can significantly alter the path the genome takes to manifest itself.

Consider also that the genomic sequences between a human and a chimpanzee differ only by 1–2% but at least one study reported that 80% of proteins are different between humans and chimpanzees (19–20). What appear to be subtle differences in genomic sequences between a human and a chimpanzee give rise to completely different species, of different physical and cognitive profiles, and different sensitivities to different pathogens.

The existence of epigenome (see box 1 for glossary) also argues against a simple relationship between the genome and the phenotype.

The genome is linear and static in the sense that it’s message is embedded in its sequence and it’s constitution remains the same throughout an individual’s life (except for when there is a tumour formation where somatic changes take place in the cancerous genome), but its expression is neither static, nor linear.

Between the genome and the phenotype, there are intermediates that bore much higher correlation with functional consequences that define who we are, and our health and illnesses (Fig. 2). These intermediates interact in networks, pathways and cascades intracellularly, intercellularly, and with the environment outside the organism. They are highly dynamic and responsive, yet they are not chaotic when we are in good health. There are spatial and temporal regulations throughout an individual’s life.

Thus trying to find a linear correlation between genome type and phenotype is not likely to yield clinically useful data except for rare variants of high penetrance, in mono- or oligogenic disorders and in familial/inbred population GWAS.

In other words, personalized medicine is far more complex than medicine based on genomic sequences and variants obtained at affordable prices. The static genome will have to be interpreted in the context of the intermediate expressionomes [all derivatives of the genome, including transcriptomes, regulatoromes, epigenomes,
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<th>Text box 1: Glossary used in the text and/or figures</th>
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<td>Genome: the sum of a cell’s or an organism’s hereditary materials.</td>
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<td>Gene: stretches of sequences in the genome that encode proteins.</td>
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<td>Gene expression: the process and/or result of a gene being transcribed into RNA.</td>
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<td>Transcriptome: the sum of all transcripts, i.e. all RNA, but may refer to only the sum of all mRNAs, in a cell or a sample.</td>
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<td>Proteome: the sum of all proteins in a cell or a sample.</td>
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<td>Metabolome: the sum of small-molecule metabolites in an organism or a sample, and include endogenous (produced by the body) and exogenous (from environment, e.g. food) metabolites.</td>
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<td>Epigenetics: the phenomenon of heritable changes in genes’ functions that is attributable to parent-of-origin (at the cellular or organism levels) instead of a change in genomic sequences. The mechanisms involved include methylation of the cytosine and post-translational modifications of histones, that change the chromosome territory and configuration, whereby influencing gene expression.</td>
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<tr>
<td>Epigenome: primarily the study of genome methylation patterns, may also include studies of histone modifications.</td>
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<td>Regulatorome: the sum of DNA binding elements and genomic sequence elements that regulate gene expressions. Examples include promoters, enhancers, miRNAs and proteins.</td>
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<td>Metagenome: uncultured and unamplified hereditary materials from an environment, e.g. DNA from the germs in the human gut.</td>
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<td>System biology: inter-disciplinary study of complex biological interactions (e.g. metabolic pathways) at multiple levels, e.g. metabolome, transcriptome and genome.</td>
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<td>Genetomics: genetics and genomics and its derivative – omics, including transcriptome, regulatorome, exonome, proteome, metabolome and epigenome, etc.</td>
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<tr>
<td>Expressionomes: intermediates between the genome and the phenotype, including transcriptome, regulatorome, exonome, proteome, metabolome and epigenome.</td>
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<td>Haplotype: 1half of the genome; 2genomic DNA sequences in cis-relation to each other (i.e. on the same molecule, as opposed to on separate molecule, as in trans); 3closely linked cis-elements in the genome that tend to be inherited together, not separated by recombination events.</td>
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<td>aCGH: array comparative genomic hybridization: whole-genome scan for microdeletions/duplications and other copy number variants.</td>
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<td>SNP: single nucleotide polymorphism.</td>
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<td>GWAS: genome-wide association studies. Usually SNP arrays are employed for GWAS. SNPs in persons with a disease, e.g. type 2 diabetes, are compared with SNPs of persons without the disease, and those that appear significantly more often in the disease group are scored on the strength of their association with the disease, often expressed as odds ratio.</td>
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This is not to say that genome sequencing and GWAS are not worthwhile pursuits. On the contrary, one cannot over estimate the value of understanding our genome. From clinical standpoint, such knowledge can lead to:

1. Better understanding of important biological questions across species as the genome language is universal on earth.
2. More reliable and precise disease diagnosis, categorization and taxonomy. For example, cancers will not only be categorized by their anatomic location and histology as lung
cancer, lymphoma, and sarcoma, but also by their underlying molecular etiology. For example, ALK gene has been shown to be involved in anaplastic large cell lymphoma, myofibroblastic sarcoma, non-small cell lung cancer, and neuroblastoma, so these malignancies maybe grouped under ALKomas (22–23).

(3) The discovery of new, more effective and more precise targets for disease intervention, with fewer side effects. Case in point, lung cancers are of heterogeneous molecular etiologies and as such they do not respond equally to the same treatment. However, ALKomas, regardless of their anatomic location and histology, maybe responsive to the same or similar treatment strategies as they share the same molecular pathogenetic etiology.

(4) More precise, reliable and efficacious tests for high penetrant genomic changes associated with clinical diseases.

(5) Increased awareness of genome-environment interactions in health and diseases, thus promoting informed and proactive public health policies and individual health choices, and transforming healthcare from doctors treating diseases reactively to consumers and societies participating in disease prevention and health promotion.

But the intermediates, the results of complex interactions of gene–gene, gene–regulatory elements and gene–environment as well as chromosome configurations, are far more likely to yield high predictive and diagnostic values (e.g. glucose tolerance test vs GWAS) and targets for therapeutic interventions (e.g. treatments for phenylketonuria), and their utilization does not hinge on our deciphering the remaining 95% of the genome.

However, in order to tackle the complexity of expressionomes in real time at cellular, tissue and organism levels, with comparable throughput as genome typing, technological breakthroughs are needed for newer and more powerful tools. Systematic studies of the intermediate expressionomes are much harder to tackle because, unlike DNAs that are made up of four bases, their chemical compositions are much more complex. The
Table 1. A simplified comparison of clinical genetics and the anticipated genomic medicine

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<th>Clinical genetics</th>
<th>Genomic medicine</th>
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<td>Emphasis</td>
<td>Rare or very rare diseases</td>
<td>Common complex diseases</td>
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<td>Genes involved</td>
<td>Monogenic or oligogenic</td>
<td>Often unknown</td>
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<tr>
<td>Genomic changes</td>
<td>Chromosome rearrangements, aneuploidy, copy number variants, deletions, duplications</td>
<td>Multiple variants/poly-morphisms</td>
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<td>Penetrance</td>
<td>High Diagnostic, carrier testing and pre-symptomatic testing</td>
<td>Low Predictive risk assessment</td>
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<td>Disease detection</td>
<td>High predictive value</td>
<td>Low predictive value (not yet proven)</td>
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<tr>
<td>Pre-symptomatic testing</td>
<td>High predictive value</td>
<td>Low predictive value (not yet proven)</td>
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<td>Treatment</td>
<td>Variable approaches</td>
<td>Personalized approach based on genome info</td>
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<td>Clinical geneticist</td>
<td>Medical doctors, well educated and trained in this specialty</td>
<td>Minimal education and training</td>
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<tr>
<td>Laboratory geneticist</td>
<td>Usually PhDs, well educated and trained overseeing clinical service laboratories</td>
<td>Usually PhD researchers without direct clinical involvement</td>
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human genome, with over 20,000 genes, produces far more than 20,000 RNA species and proteins through regulated expression, alternative splicing and post-translational modifications (24). And unlike DNAs that exist in cells in diploid sets (except for mitochondrial DNA), the amount of each component in the expressionome can differ by up to tens of thousands or millions of fold. So despite mesmerizing technological advances, the distance to cover is great. Case in point, a few decades ago, researchers’ abilities to generate monoclonal antibodies elicited quite some excitement as it promised ‘magic bullet’ therapy for cancer and other disease conditions. Although progress has been made, to date, only less than a handful are in use for cancer treatment (25–27), still fewer with undisputed efficacy, although admittedly more of them are in use for clinical tests, and still more for laboratory research.

On the other hand, one has reason to be optimistic that where limited tools are available, even small scale data generation in expressionomes potentially can be translated into clinical utility much faster than genomic data. For example, CA-125 as an indicator for ovarian cancer has a diagnostic sensitivity of <60%, but a multiplex immunoassay using six markers (leptin, prolactin, osteopontin, insulin-like growth factor II and macrophage inhibitory factor together with CA-125) achieved a 95.3% diagnostic sensitivity and a 99.4% specificity (28). Similar levels of sensitivity and specificity have also been achieved on protein microarrays for severe acute respiratory syndrome (SARS) (29).

Just as our limited knowledge of the space has not precluded our launching satellites to facilitate communication and weather forecast, our ever expanding knowledge of the genetomes (genes, genome and – omes), although incomplete and emerging, can be harnessed to benefit healthcare. Alongside these progresses, there is a want for regulatory guidance on how to incorporate such advances into medical practice, or perhaps more immediately, how to protect consumers of genetomic healthcare. Moreover, clinical utility involves not only the demonstration that genetomic studies are of clear benefit, it is also related to consumer’s understanding and acceptance of such tests and interventions, and the downstream impact on the consumer’s behaviours and its measurable positive health outcome.

The societal ethical and legal aspects of utilizing genomic data

Regulatory bodies can impose standards for diagnostic laboratories to meet (see box 3 for a few government and societal initiatives on the regulation and promotion of genomic personalized medicine). The need for such standard-upholding regulations has attracted a lot of attention and debate, in particular, in response to direct-to-consumer marketing of genomic medicine and recreational genomics (30, 31). The alarm bell went off not only because these commercial companies are bypassing healthcare professionals, but also because they are offering tests of unproven clinical validity and/or utility. It is thus feared that personalized medicine could become alternative medicine, only this time, massive personal information is at stake. However, regulating these commercial laboratories and their standards is not
Text box 2: The human genome project and some of its derivative projects and initiatives

The human genome project: http://www.ornl.gov/sci/techresources/Human_Genome/home.shtml

The 1000 genome project: a project to sequence 1000 human genomes to be used as references. http://www.1000genomes.org/page.php

HapMap: a project to compile human haplotypes with the goal of identifying and cataloguing similarities and differences in human genomes. http://snp.cshl.org/

GWAS catalog: up-to-date cataloguing of all GWAS studies. http://www.genome.gov/26525384

Cancer genome atlas (TCGA): a project to catalogue genomic changes in various cancers that lead to the cell’s cancerous transformation. http://cancergenome.nih.gov/

Gene Expression Omnibus an online expression data repository for the storage and retrieval of gene expression data from any organism or artificial source. http://www.ncbi.nlm.nih.gov/geo/

MGED, the Microarray Gene Expression Database. http://www.mged.org/

Micro Array Markup Language (MAML): a standard platform for laboratories worldwide to submit and perform comparative analysis of microarray expression data.

Genome 10K: collection of DNA sequences representing the genomes of 10,000 vertebrate species, approximately one for every vertebrate genus. http://www.genome10k.org/

The human microbiome project (HMP): comprehensive characterization of the human microbiota (e.g. gut flora) and analysis of its role in human health and disease. http://commonfund.nih.gov/hmp/


Text box 3: A few government and societal initiatives on the regulation and promotion of genomic personalized medicine


U.S. Congress S. 976 Title: A bill to secure the promise of personalized medicine for all Americans by expanding and accelerating genomics research and initiatives to improve the accuracy of disease diagnosis, increase the safety of drugs, and identify novel treatments.


Genetic Information Non-discrimination Act (GINA): http://www.genome.gov/24519851
the only issue surrounding genomic medicine, and probably not the most complicated either.

A person’s genome and expressionomes hold secrets to an individual’s health and illnesses, as well as his/her intelligence, personality, sense of humour, curiosity and so on. Indeed, since 2007 curious people have turned to commercial companies to have their genome scanned for a price starting from $399 (https://www.23andme.com; http://www.navigenetics.com). A Swiss company GenePartner even offers to help people to find the perfect romantic match for just $99 (http://genepartner.com). Such practices are dubbed ‘direct-to-consumer’ (DTC) genomic testing.

Google (http://www.google.com) has also launched Google health with the aim of enabling individuals to store their personal genomic and health information online so that relevant info can be retrieved through Google search in the future. Such storage of huge amounts of personal genomic information is significantly beyond direct-to-consumer testing, it is really the essence of consumer-directed healthcare (CDH). When commercial companies can offer, at the request of the consumer, testing of the static genome and its dynamic expressionomes at any point of care, or curiosity, several questions beg to be addressed.

(1) Should the Google and similar endeavours be permitted or forbidden? While the public and individuals have the right to know what they want to know, information that is incomprehensible, incomplete, or inaccurate could lead to misunderstanding and potentially adverse consequences. Standard-upholding regulations are indeed needed.

(2) Who owns an individual’s genomic information, the individual and his/her legal executor or medical power of attorney, the spouse/co-habitant or biological next of kin, the healthcare facilities, the payer for healthcare (employer, insurance company, government/tax payer) or the companies that collect, analyse, store and retrieve the data?

Ownership directly impact the individual’s right to maintain or destroy their genomic information, their wish to contribute or not to contribute their information to any public or private databases, and their participation, access and utilization of results from familial GWAS, and so on. If expectant parents own the info for their pregnancies, what stops them from trying to have the designer baby? If parents knew their newborn baby has high risk for schizophrenia or violence or low IQ, would the baby be loved and cared for the same way when such info was not available? Should potential adoptive families be made aware of such info prior to their decision making?

The significance of 95% of the genome is unknown, so people who are having their genome scanned literally do not know what they are getting into and the emerging knowledge, incomplete as they are, at a later time may put a label or deliver other psychosocial impact the person is not prepared for. Moreover, the person’s children, grandchildren or great grandchildren may be living with the consequences of such information that they do not have adequate control or any control over its dissemination and usage.

If the bio-specimen and the info related to it are owned by private companies, can they be traded as commodity? If yes, how should they be regulated? What happens when there is a merger, acquisition, spin-off, bankruptcy? What happens when multiple companies are involved in the process of data collection, storage, retrieving?

If such info is owned by healthcare facilities, can an individual control who has access to it and when? For example, if a 50-year-old man is involved in a serious motor vehicle accident (MVA), and his genome info shows he has a 85% lifetime risk of developing early onset Alzheimer disease, would and should the emergency physician treat him the same way as someone with a very low risk for Alzheimer disease in his lifetime? What if, instead of MVA, this is a case of prostate cancer or knee replacement or coronary bypass surgery?

If the payer owns such info, and in many systems there are multiple payers, should they turn a blind eye to such info which may have tremendous implications for healthcare cost, Genetic Information Nondiscrimination Act (see box 2 for a list of regulatory bodies and their guidelines) notwithstanding?

If the government owns such info, should law enforcement keep a tag on the person whose genomic info indicates a high predisposition to violence? And in the case of an unsolved violent crime, should law enforcement be allowed to search databases for individuals of such predilection? For suspects of a violent crime, should their genomic info be used as an additional piece of evidence against them in the court of law or to exonerate them because they have no control over their genomic inheritance?

(3) Who should be at the rein for personalized healthcare, the consumer, the commercial companies, the doctors and healthcare facilities, or the payers? Some doctors are reluctant
to operate on obese patients or smokers due to high peri-operative complications. In the future, when a patient comes for surgery, or any other treatment, say even a simple wart removal, can the doctor demand that the patient go through genomic profiling or sign a consent to release all his/her genomic info or swipe his/her smart health card before any treatment is rendered? Should the payer cover genomic profiling for such trivial purposes as a wart removal? And in the event of an adverse outcome, could the doctor be sued on the basis of not having reviewed the patient’s whole-genome info? Further, if endeavours such as Google’s lead to a consumer-directed healthcare through direct-to-consumer testing and data retrieving, would it supplant the traditional physician-patient relationship? How should accountability be distributed?

If personalized medicine is largely about risk assessment and predictive/preventative intervention, how would the healthcare system and the insurance industry pay for such tests and preventative interventions when a disease condition is not existent? If such claims for healthcare and insurance coverage are valid, then discrimination based on a person’s genomic information will become inevitable. If they are not covered or covered differentially depending on one’s policy, then justice as it relates to access to healthcare could be compromised. Admittedly, access is an age-old issue, but as we enter into the new era, the gap between haves and have-nots can become more divisive.

The genetic code is universal and a person’s genome is static. So unlike active medical diagnosis and management where real time assessment is mandatory, be it in person or through tele-health or e-health, genomic predictive info can be interpreted and communicated to the consumer without borders, without a patient–physician relationship and at all hours. Can the reading of genome data output be outsourced to other countries where regulations are different or lacking? Can one medical expert in New York render his opinion directly to a consumer (as opposed to acting as a consultant to the consumer’s healthcare provider) in Hawaii or Dubai, if the consumer pays for it? How would this affect malpractice insurance and/or licensing?

Another dimension of personalized medicine is the concept of individualized drug treatment, also known as pharmacogenetics and pharmacogenomics. Currently, the development of new drugs involves virtual and/or laboratory design and in vitro testing, followed by animal and/or ex vivo testing, and then clinical trials, typically of phase I through phase III. Such process primarily looks for drugs that are safe and effective for the mass so that production and marketing eventually generate a handsome profit.

While on one hand, genomic information could potentially reduce the time and cost of drug design and trials by improving their predictive value and through pre-identified subsets of participants in the trials, such reduction in cost may not offset the length of time to generate a profit on a much smaller user volume. Indeed, the cost of clinical trials may go up instead of down if the company has to recruit participants across communities or countries in order to have adequate sample size for a particular genome type. Would this mean that people with rare genotypes or rare combination profiles could be differentially neglected in such drug development model? Or would they be paying a much higher premium for a medication thus developed? Would this then lead to a higher premium for drug/health insurance and consequent discrimination by insurance companies and other payers?

How should genomic information guide clinical management to ensure that no harm is done? Autopsy studies have shown that the actual incidence of cancer was far greater than clinically recognized, suggesting that a healthy person’s immune system could conquer many occult cancers in the person’s lifetime (32). So how should one’s genomic profiling be used to guide decisions on preventative and therapeutic intervention without over treating pre-clinical cancers and other conditions? How should potential conflict of interest of commercial companies that offer predictive testing and intervention be regulated?

All these yet-to-be resolved issues notwithstanding, a paradigm shift (Fig. 3) in healthcare, driven by the trio of research, commercial companies and consumers, is gaining momentum not unlike the internet two decades ago, however unprepared the regulatory bodies are. Geneticists, as professional healthcare providers, should team up with researchers, educators and other professionals involved in healthcare to lead this change, to inform policy-makers and to provide professional guidance that addresses the needs of consumers.
J. The regulation of direct-to-consumer genetic tests.


32. Thompson IM, Lucia MS, Tangen CM. Commentary: the ubiquity of prostate cancer: echoes of the past, implications for the present: “what has been will be again, what has been done will be done again; there is nothing new under the sun.” ECCLESIASTES 1:9. Int J Epidemiol 2007: 36: 287–289.

Fig. 3. Paradigm shift in healthcare. dz: disease.

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References


