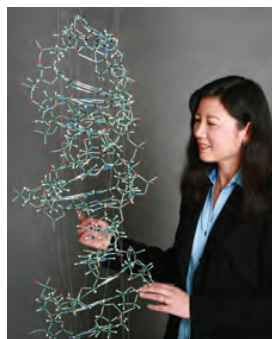


The Advent of Consumer Genomics

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The science of genomics is swiftly developing into a powerful tool for making personal decisions that can improve our health and quality of life. The rapid evolution of this technology is being led by the convergence of two ongoing technological transformations, the growing knowledge of the links between genetic variation to health and wellness, and the falling cost of genotyping and sequencing. This convergence is giving us individualized knowledge of how our genetic makeup predisposes ourselves to a host of medical conditions and shapes our behavior.

Further, the genomic technology breakthroughs occur at a time that internet has transformed how information and service are delivered. An increasing number of people go to the web first for healthcare information and want more control and convenience over their individual healthcare decisions. In contrast to traditional patients who receive treatments prescribed by their doctors and as dictated by medical insurance, consumers are increasingly expecting more control and options in their health care. They increasingly seek preventive care before they get sick, choose services that suit their personal preferences and convenience, and in general are taking more control of their well-beings. In the expanding field of genomics, consumers- rather than the traditional medical providers- may lead the way by first accessing genomic technology on their own then sharing their genetic information with healthcare providers on an as-needed basis.

Thus born from the large scale private and public genomic efforts of the last decade, genomics is coming out of the research lab and into the consumer market. The field of consumer genomics is promising to create exciting business opportunities which might lead to radical changes in how healthcare service is delivered. Consumer genomics has the potential to fundamentally improve how we approach personal health decisions, as well as making life more fun, relaxing and enjoyable.

Consumer genomics and personalized health

Personalized medicine is often defined as delivering the right drug to the right patient at the right time. It can be more broadly defined as personalized health, the ability for us to make lifestyle and medical decisions based on our individual nature. Though long discussed, personalized medicine has been difficult to deliver. Consumer genomics creates the means to deliver many of the promises of personalized medicine on a scale and cost which was not possible before. With the potential advantages of improving personal health, lowering health care costs, and creating novel market opportunities for health care providers, it is expected that the demand for consumer genomics will be driven on multiple levels, and will soon be an important element of personal health care decisions.

In these early days of consumer genomics and personalized medicine, people can already use genetic information to make a growing number of health care decisions. Pharmacogenomics has identified a number of genetic variants that can be important in determining treatments and drug dosages that are effective yet with the least risk of side effects. Several companies offer direct-to-consumer genetic testing, and traditional medical providers are also becoming more comfortable using

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this novel information to guide treatment decisions. These companies offer genetic counseling services or other decision making tools and interpretation aids, allowing consumers to make informed choices and understand their test results.

Our DNA also shed lights on more than just our health, and an understanding of our personal genome will allow us to comprehend ourselves in a vitally new way. Genetics help us to explore our ancestry. It also tells much about our individual traits, from our personality, athletic ability, to how we taste food, smell perfume and experience music. All these facets of ourselves have genetic components that we share among our family members and ancestral groups. Personal genomics offers the ability to explore these genetic relationships for the first time. Of course, it is also important to note that while our genes help shape us, they are far from the only determinant of who we are.

Technological transformations lead consumer genomics

Primarily the genetic tests offered to date are focused on at most a few genetic variants that map to a single enzyme or biomarker. Moving beyond single mutation analysis, the new technologies that promise to dramatically widen the impact of consumer genomics are high density single nucleotide polymorphisms (SNPs) chips and whole genome sequencing. The Affymetrix Genome-Wide Human SNP Array 6.0 is a recent example. This chip detects over 906,600 SNPs and more than 946,000 copy number variants. Illumina also offers a competing SNP chip technology, the Human1M DNA Analysis BeadChip, featuring more than one million SNPs for genotyping and copy number variant analysis.

The cost of DNA sequencing continues to drop, and a variety of techniques promise to make sequencing the entire genome, or at least the ‘exome’ (the coding sequence of all proteins) comparable with genome-wide SNP analyses. Several companies and academic efforts are developing sequencing technologies, and the \$10 million ‘X-Prize’ is to be awarded to the first team that can sequence 100 human genomes within 10 days for a cost of no more than \$10,000 per genome. The 454 Life Sciences, Microchip Biotechnology, Applied Biosystems, the privately funded Personal Genome Project, and several other groups are actively pursuing novel sequencing technologies. Though the methods differ, all efforts use microchips to allow for miniaturized and highly parallel reactions. If the costs of these techniques can continue to fall, the greater information content provided by whole genome sequencing may make this the dominant technology of the consumer genomics field.

As important to consumer genomics as the technologies allowing whole genome information to be quickly and cheaply acquired is the need to be able to relate genetic variation with personal medical decisions. Fortunately the unraveling of complex diseases is being rapidly propelled by the growing sophistication and feasibility of genome-wide association studies (GWAS). In 2007 alone there were more than 20 GWAS, each providing new information about how genetic variation can be used to predict disease risk and treatment outcome. Though new technology is making these studies increasingly economical, the scale of these efforts is still daunting and collaborations are proving important in increasing the resources available for these surveys. An example is the Genetic Association Information Network, GAIN, a partnership between the pharmaceutical industry, the National Institutes of Health, and others to share genome-wide association data. As GWAS continue to accumulate data, and the ability to share and compare this information increases in sophistication, it is expected that the amount of information in our genomes that can be used to guide our choices about personal health will rapidly grow.

Personal genomics enters the commercial field

A number of companies have been recently launched bring genetic technology and information directly to consumers. These companies make it possible for people to obtain their personal genetic information directly without having to rely on traditional medical providers to provide this information. It is likely that the established medical portals will lag considerably behind these new companies in providing genomic services. Most of these companies are internet-based with, as yet, no physical storefronts. Customers generally provide their own DNA via a cheek swab or saliva collection kit and the consumer genomics company handles the DNA extraction, analysis, and reporting.

23andMe is a web-based service using Illumina's bead-based SNP detection technology. Their current marketing emphasizes the excitement of using this novel approach to understand yourself. Besides medical decisions, *23andMe* promotes the use of personal genomic information as a tool for exploring one's ancestry, sharing of family traits, and generally exploring the information contained within one's genomic variation.

deCODE Genetics is positioning themselves similarly to *23andMe*, stressing the personal exploration aspects of personal genomics as much as the medical aspects. However, *deCode*'s proprietary information resulting from its large scale population studies may give it some advantage in providing relevant medical associations.

Navigenics offers a whole genome scan with the Affymetrix 6.0 chip, and stresses the medical advantages of whole genome scans more than *23andMe* or *deCODE*. Their service employs a personal web account to relate the personal genomic information to relevant health and wellness information, consultation with genetic counselors and a subscription to allow one to compare their genomic information with new developments in genetics and treatment.

DNA Direct pioneers direct-to-customer DNA testing and online medical genetic service. It provides consultation service to those considering or already having performed a whole genome scan, helping clients understand their test results and how to act upon them. It also offers a broad range of consumer-friendly tests and services for specific disorders and traits.

Knome is the first privately funded, for-profit company offering whole genome sequencing. Their initial efforts will be limited to 20 individuals, with prices starting at \$350,000, including whole-genome sequencing and analysis from a team of geneticists, clinicians and bioinformaticians.

Personal Genome Project is a not-for-profit effort led by George Church of Harvard University. The goals are to explore the most effective means of sequencing and communicating personal genomics information in an economical, effective, secure, and ethical manner. Their initial efforts are focusing on ten volunteers and sequencing the exome, with plans to scale to 100,000 volunteers in the next several years.

Prospects for consumer genomics in china

Chinese scientists have played integral and significant roles in the successful completion of the Human Genome Project in 2001 and the International Haplotype Project in 2005. In October 2007, Chinese scientists announced the completion

of the first sequence map of a Han Chinese individual, which represents the third human genome to have been sequenced in the world. This Chinese project was led by the Shenzhen branch of the Beijing Genomics Institute. The institute announced that the next step of the project will be to sequence the genomes of more individuals to identify genetic variations in Asian populations and explore the essential mechanisms behind many diseases and traits. The researchers will soon select 99 Chinese people for the project, and the number of research subjects will be expanded to 10,000 in the following few years. Similar large scale research efforts on Asian populations are also underway in several other genome centers and academic institutes. The availability of such information in Chinese population, along with the international efforts to share and compare the whole genome association study data, accelerates the accumulation of knowledge on our genomes that can be used to guide our choices about personal health.

Consumer genomics is likely to grow fast in the Chinese market. Chinese consumers are rapidly gaining greater wealth and are willing to pay for better health and wellness services. The payers in China, including both HMOs and high-end service plan, are highly entrepreneurial and are willing to entertain new business models employing the latest technologies. The high productivity and deep talent pool for both laboratory and informatics personnel in China will enable China to be a highly competitive global genomics service center. Most importantly, the entrepreneurial spirit and environment in the Chinese life science sector is astounding and China has been the fertile ground for many emerging innovative companies. At this early stage of consumer genomics, although it is not clear how consumers will embrace genomics while the issues about regulation, privacy, payment and business models remain to be worked out, there is no doubt that genomic technology will prove to be an invaluable tool to improve people's health and life.