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## Genomic Data in Patients' Hands

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Strides in biotechnology and genomics have enabled individuals to gain access to increasingly more accurate and detailed information about their genomes at decreasing costs.

While learning about individual genetic risks and susceptibilities has become more affordable, interpreting the data and establishing the optimal management strategies are opening challenges for both consumers and professionals, and many of these promise to become more pronounced as we approach the \$100 genome landmark.

"As a molecular biologist, I was driven by how fast advances in genome sequencing are delivering information, and by the gap that was created and still exists in clinicians' ability to understand this type of data," says Patrick Merel, Ph.D., founder of Portable Genomics. Established last year, the firm proposed to provide a user interface to help clinicians and consumers interpret our genome data simply and without the need for an advanced degree in molecular biology.

"We came with the idea to bring digital media interfaces into genomics and organize the genome data as digital music files," says Dr. Merel. Portable Genomics mostly works with datasets provided by 23andMe, but it anticipates using full-genome data from any other provider in the near future. The list of traits that is provided after genotyping can be downloaded as a large file of raw data that contains a list of SNPs.

"We rearrange full-genome data about diseases and traits into an mp3 structure, and provide a file that can be read by iTunes or any similar software," explains Dr. Merel. The data files can be stored and shared as digital music, on various types of devices, including mp3 players, iPods, and digital TVs, and even cloud-based digital music services, such as Amazon Cloud Player, Google Play, or iTunes Match.

While it is often challenging to bring computers with 3 GB datasets to a consultation, the availability of the data on a portable device, such as a smart phone, which is supposed to have an mp3 player embedded, allows patients to share the information with healthcare providers, and a physician can rapidly sift through high-risk traits.

"By early 2015, we anticipate providing a subscription-based mobile application for the full genome for approximately \$100 a year," says Dr. Merel. This visualization tool will facilitate the incorporation of genomic information into prevention strategies. "We have designed a few functionalities to help patients learn about their risks and susceptibilities and allow them to take action," explains Dr. Merel.

For example, the application can use location information to help a patient who learned about a heightened risk for breast cancer promptly identify a specialist nearby. For every trait that is reported, a function will remind patients to schedule appointments, and will do so until an appointment is made.

"If we want to help patients use genomic information for prevention, we need this kind of application, because we know that we cannot rely on people spending time to examine their genetic risks and explore the course of action," says Dr. Merel. Additionally, real-time updates will incorporate newly emerging links between mutations and risk factors. "Genomics will be such a moving field during the next 25 years that access to real-time monitoring is crucial," emphasizes Dr. Merel.

### Set Your Genes to Music

An additional line of work at Portable Genomics, based on this digital media approach, is related to the idea of making music from our genome. "This concept is not something new, there are many projects that generate music from DNA sequences, but what is different is that we are bringing in the use of personal genomics for entertainment," says Dr. Merel.

An initial concern was the possibility to record the music and decipher it back into genomic information, but an algorithm developed by Dr. Merel and colleagues, which involved encoding the genomic information into a numeric key, helped address this issue.

"From the numeric key, a key that is representative of a genome, one cannot go back to the genomic data, and this allows people to play with and share the data without any privacy issues," says Dr. Merel. He and colleagues incorporated this algorithm into GeneGroove, an iPhone application that downloads 23andMe raw data, generates the numeric key, erases the genomic data, and plays a unique tune from that key.

"It seems that genomics also found a way to serious gaming applications, and can stir interest for and raise awareness

about the genome among the public,” notes Dr. Merel. This endeavor has already received much interest from the music industry. “We are working with major artists who have been intrigued by using their genome as a way of creation, and we found that this can impact interest among consumers for learning about their genomes,” says Dr. Merel.

### **Genetics vs. Lifestyle**

“Our mobile application provides a great opportunity and a great medium for people to bring their genomic data to their physicians, and this is particularly important with regard to drug response results,” says Mike Polcari, head of software engineering at 23andMe.

As part of the personalized genomic services that 23andMe offers from a saliva-based test, over one million data points, obtained for every person, are analyzed and help generate approximately 246 different reports. These include disease risk reports for conditions such as Crohn’s disease, celiac disease, types 1 and 2 diabetes, Parkinson’s and Alzheimer’s disease, reports on physical traits, a carrier status panel, and information about pharmacogenetics and genetic ancestry.

“Many of these conditions are shaped by other factors, such as lifestyle, and we make significant efforts to be very clear that our reports are informative only about the percentage of risk that is attributable to genetics,” says Catherine Afarian, head of public relations at 23andMe. The mobile application released by 23andMe, free for download, is currently available for Android and iOS.

“We found that people responded very well and have also been participating in research on the mobile application in large numbers,” says Polcari. Most of the high-quality health and trait reports are currently available on this mobile application. 23andMe uses a star rating system to prioritize genetic risk factors, based on the body of literature published on a specific link.

“We have four-star reports for links that are supported by more studies and have therefore been accepted by the scientific community, and we prioritize and put those onto our mobile applications first,” says Afarian. For other genetic risk factors, one-, two-, or three-star reports reflect the fact that fewer studies were performed to date, and further research to replicate and validate those findings is required before placing as much confidence in them. “As new research is being done, we are constantly providing new reports to our customers,” adds Afarian.

### **Reconciling Mismatched Information**

Services from distinct companies sometimes provide different pieces of advice, and this may be a source of confusion for both consumers and professionals.

“In 2009, I designed a mobile application platform to provide a side-by-side comparison of different genes and SNPs that different consumer genomics providers review,” says Melanie Swan, founder of DIYGenomics, a nonprofit research organization that proposes to use genomic information to complement the information consumers obtain from healthcare professionals.

A free and open-source application offered by DIYGenomics, currently informative on up to 20 health conditions, the response to 200 different drugs, and athletic performance information, is available on three platforms: Android, iOS, and Windows Phone. “We’ve had about 10,000 downloads,” says Swan.

Genomic information uploaded on the mobile application can actively guide healthcare decisions. For example, a patient with heightened risk for a specific cancer may decide to undergo more frequent screenings, or use information about athletic performance to customize training programs. “In my case, I have more mutations in my strength-related genes, and this prompted me to include more weight-lifting into my daily regimen,” reveals Swan.

These developments are paving the way for the time when personal genome information will be included in everybody’s medical file. “We are coming into an era where health becomes an information science, and by integrating multiple different types of data files about an individual, we can use quantitative data at the level of risk and probability to develop a preventive medicine strategy and work toward keeping people healthy,” explains Swan.

### **Exploiting Cell Phones’ Reach**

“We wanted to develop a portable device to perform DNA- and RNA-based assays,” says Syed A. Hashsham, Ph.D., professor of civil and environmental engineering and Michigan State University. Recently, Dr. Hashsham and colleagues described Gene-Z, a user-friendly device that relies on a disposable microfluidic chip, and allows the simultaneous quantitative detection of multiple genetic markers with high sensitivity and specificity.

In this application, real-time measurement of isothermal amplification products using fluorescence allows the quantification of multiple genetic markers at a single temperature, and the wireless interface and smartphone application for data analysis and reporting illustrate its utility for affordable and reliable point-of-care genetic testing. Point-of-care tests are ideal for assays that have immediate or long-term health benefits or may save lives, and for those that present a minimal risk for false-positive results and have full support systems in place for consumers undergoing testing.

“So there may be some assays that are never good for a cell phone, while other are very good,” says Dr. Hashsham. This strategy is not limited to genetic assays, but any device developed for genetic assays may also be adapted for

antibody-based or chemical assays, because most reactions rely on colors that develop based on a specific signature molecule.

“What I have next in mind is using color assays for diseases such as diabetes or measuring cholesterol levels,” says Dr. Hashsham. One of the challenges with point-of-care testing is not the technical challenge but the business model, because an emerging or a new technology generally boasts about its low cost but at least in the initial stages must get off the ground without high sales volumes.

“But if we are able to develop genetic diagnostic applications on cell phones, we can reach billions of people without the need for expensive monitors. As we all know, the number of people with a cell phone is already more than those with a toothbrush,” says Dr. Hashsham.

### **Real-Time Monitoring and Feedback**

According to recent Centers of Disease Control and Prevention estimates, 80% of heart disease and strokes and 40% of cancers are preventable through the elimination of risk factors. A key requirement to facilitate the development of management strategies is the identification of actionable risk factors. Mobile health applications, which involve technologies that monitor parameters such as heart rate, body weight, and blood pressure, help integrate these components, which normally would not be brought together, and allow healthcare teams to gather real-time data while the patient is not even in the office.

“All these data allow real-time continuous monitoring and, most importantly, real-time feedback,” says Samir Damani, M.D., PharmD, founder and CEO of MD Revolution. By allowing people to track their activities and increase their awareness of what they are doing, these initiatives help motivate changes in behaviors, such as healthier eating habits or increased exercising. “Mobile health is an enabler to measure, monitor, and motivate,” says Dr. Damani.

MD Revolution has its own software system that helps integrate these mobile components, and shares them not only with the medical team but also with the patient. “One of this model’s strengths, in comparison with traditional healthcare settings, is patient empowerment through mobile technology and genetic information, and this will shift the paradigm from a doctor-centric to a patient-centric model of healthcare, in this country and beyond,” explains Dr. Damani.

Applications that bring genetics and genomics closer to consumers promise to start filling a longstanding gap in public awareness and preventive medicine. These platforms help identify medically actionable information and deliver strategies for prophylaxis, therapy, and surveillance, and their development is foreshadowing a time when consumers and healthcare teams can routinely incorporate personalized genomics into lifestyle decisions.