SEQUENCING YOUR GENOME: IT CAN HAPPEN TO YOU, TOO!

Genome Sequencing is a way of “reading” DNA molecules — two strands twisted together to form that famous double helix. The entire human genome contains roughly 3 billion molecular base pairs, which researchers study to find variations that may play a role in the origin and development of disease.

Rapid advances in the technologies that give scientists the ability to analyze, understand and identify the unique characteristics in the genome of every human being, are now being translated into clinical applications that prolong the life of many individuals afflicted with a variety of diseases. This was evident at the 10th anniversary of the Personalized Medicine Conference, sponsored by Harvard Medical School and Partners Healthcare, last week in Boston.

The presentations focused on the great strides that have been made since the conference began a decade ago, when conference participants were talking only about the possibilities that genome sequencing might offer, along with the high costs associated with this technology.

It was evident this year that those possibilities have evolved into concrete clinical applications, and the costs have dropped to a manageable level. Much of the discussion centered on some of the ways that sequencing can be critical in addressing many diseases, particularly in the field of oncology. That these proactive clinical applications are taking place and saving lives is remarkable and makes it perfectly clear, that what was considered almost impossible as recently as five years ago, is happening .

The cost of sequencing, (a much discussed topic over the past ten years) which was a prohibiting factor just a few years ago, is also lining up to make this technology not only feasible but realistic. When one of the first individual genomes was sequenced in 2007 — that of James Watson, co-discoverer of DNA’s double-helix shape it cost around $1 million.

According to an article in Nature, it currently costs $1,000 to $4,000 to map out an individual's genome. (Specialized sequencing — for, say, a cancer patient – might be more expensive.)

Ultimately, the goal is to bring the cost down to @ $100 and to screen each infant at birth to see if they have any of the known genetic conditions, or are a carrier, so that potential diseases can be addressed early on. Although it will probably be several years before this process for all newborns is accepted and feasible, it will happen.

While almost all diseases have a genetic component, it is still not entirely clear how the information obtained from sequencing the genome will translate into improved care. There are methods, even today, to find the biomarkers that identify irregularities in the genome, that enable physicians to make alterations that help remediate the problem.

Incorporating genetic knowledge and genomic technology into clinical medicine to empower clinicians with tools that predict susceptibility to common diseases and determine the prognosis for those diagnosed with a particular ailment, requires an enormous amount of data analysis. Right now, the process that would enable the clinician in a busy practice to review these huge data sets and draw conclusions that would help their patients, is not evident. There is a huge education process that has to take place, before we move from genome sequencing and analysis in the research environment to the point where clinicians can easily receive, understand, and extract the specific data to address disease implications and make informed treatment choices for each individual patient.

That day will come, but it comes with many hurdles and challenges. These include:

1. Is this yet another way that will divide the population into those who can afford sequencing and those who cannot? (e.g. The digital divide)
2. Will this work be covered by health insurance?
3. How do we find a solution to managing the massive data sets that come from sequencing the human genome?
4. How will this data be stored and regulated over time?
5. What are the privacy issues, and personal employment risks

associated with genome sequencing and how will they be addressed?
6. What are the ethical considerations when researchers start tampering with genes to correct legitimate disease concerns, and that tampering leads to more insidious behaviors such as creating a master race?
The hope is that along with the rapid strides that were discussed at the Personalized Medicine Conference for addressing serious disease, we also focus on these legal and ethical challenges before we get too far down the path of making this multidimensional technology more ubiquitous.
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UN FORUM INCLUDES HEALTH IT RECOMMENDATIONS

The Commission on the Status of Women (CSW) was established by the United Nations Economic and Social Council (ECOSOC) in 1946. It is the principal global policy-making body dedicated exclusively to gender equality and advancement of women. The active participation of non-governmental organizations (NGOs) is a critical element in the work of CSW. NGOs have been influential in shaping the current global policy framework on women's empowerment and gender equality: the Beijing Declaration and Platform for Action.
On November 3-5, at the United Nations in Geneva, I was invited to speak in the Geneva NGO Forum –Beijing +20 Un ECE Regional Review which focused on gender equality in all spheres of life. The five areas of priority in the NGO CSW agenda include:
o Women's Rights Peace and Justice
o Women's Economic Empowerment and Employment
o Displacement and Migration
o Women's Health and Education
o Violence against Women & Girls
I participated in the Women and Health interactive roundtable, representing The Global Alliance for Women's Health (GAWH) a non-governmental organization (NGO) in Special Consultative Status with the United Nations Economic and Social Council and committed to advancing women's health in all stages of life and at all policy levels,

through health promotion, education, advocacy, and program implementation.

My presentation, entitled: **Expediting Advances in Women's Health through Internet and mHealth,** focused on the three core elements of digital healthcare:

1. Full information at the point of care: the right information on the patient, in the right place, at the right time.
2. Communication, collaboration and continuity of care
3. Data access, chronic care management and population health.

I described the digital tools that support each of these core elements including:

Digital health records and health information exchange to achieve full information at the point of care;

Patient portals that enable email and e-visits for continuous communication,

Short Message Systems (SMS) that send text message notifications and reminders to smartphones and tablets;

Smartphones apps that foster collaboration between patients and providers;

Chronic care management tools such as telemonitoring systems, wearable devices, smart medication dispensers; and many of the apps that monitor body vitals and interconnect with providers for analysis and treatment decisions.

Telemedicine including video-conferencing that enables quality care to be delivered to patients who reside in remote locations where there is a shortage of health care workers.

I talked about the need for patients to have access to data and information and how that is accomplished via the Internet, where patients can search for health information and connect with social networks so that they can better understand their health issues and make better health choices for themselves and their family.

I also pointed out that no matter how isolated certain patient groups are, 98% of the world's population have a basic cellular phone, enabling them, at the very least, to receive text messages (SMS) that provide reminders about immunizations, taking medication, and

keeping appointments with health providers, nutrition advice, and more.

I concluded my presentation by pointing out how digital communication technology, the Internet and mHealth foster empowered patients – women who take charge of their health issues and manage health care for themselves and their families; engaged patients, women who actively participate in the health care experience by using digital tools to collaborate with their providers to make good health care choices and treatment decisions; educated patients who use digital tools to advance their knowledge about what is feasible and available to them.

As a result of my presentation, digital technology in health has been included, for the first time, as one of the recommendations in the outcome report from the Geneva NGO Forum, which will be part of the CSW report submitted to the UN Economic Commission for Europe for the annual CSW meeting in New York in March, 2015. At this meeting representatives of Member States will gather at United Nations Headquarters to evaluate progress on gender equality, identify challenges, set global standards and formulate policies to promote gender equality and women's empowerment worldwide.

Expediting Advances in Women's Health through Internet and MHealth.final