

Think You Know Human Genome Sequencing Pros and Cons? Think again!

The Human Genome Project (HGP) is arguably the biggest and the most ambitious project embarked upon by mankind in its scale, objectives and breakthroughs. Its genesis goes back to October 1990 when a global collaboration endeavored to decipher incisive information about the 80,000-odd human genes.

This initiative of gigantic proportions, which was sponsored by the National Human Genome Research Institute (NIH), concluded in 2003 and had a singular objective – to make human genes available for further research and development.

Why was HGP so painstakingly difficult?

Genes, which essentially defines who you are (mentally, physically, emotionally, psychologically, etc) made up of DNA, which comprises of four 'bases' or ingredients. One gene potentially comprises of thousands of bases, which implies that scientists were dealing with a massive number wherein the chronology and structure was unidentified and untraceable.

Secondly, they also needed to explore the nucleus of the cell where the DNA exists in a coiled form, uncoil it, evaluate its bases and determine their exact sequence meticulously. The importance of arriving at the correct sequence cannot be overemphasized because it practically unravels the mystery of life itself.

In other words, genes unlock the key for creating proteins, which determine everything about you and your life. This brings us to the next major component of HGP- whole genome sequencing.

Human Genome Sequencing

Following the resounding success of this venture, another far-reaching project was announced for 1998-2003 to derive the sequence of three billion odd DNA subunits wherein a number of high profile R&D activities were initiated on well-established biological models to understand the role, functionalities, and challenges of human genes.

What is Whole Genome Sequencing?

Complete genome sequencing refers to the lab-based scientific process for deducing an organism's DNA sequence at a given point in time while allowing for a thorough evaluation of any genetic variation or ambiguity. This process assumes great significance because almost any experiment involving even a tiny fraction of a DNA can pave the way for complete genome sequencing.

Whole genome sequencing services have becoming a hot topic of debate, with several companies like Illuminata Inc. and 23 & Me offering low cost genome sequencing offerings ranging between \$100 and \$4000. While many have embraced this cost lowering, others wonder as to where this hysteria would lead us.

<http://www.healthfitnessinfomagazine.com/biotech/think-you-know-human-genome-sequencing-pros-and-cons-think-again/>

Human Genome Sequencing Pros and Cons

Human genome sequence has its share of admirers and critics. Here are some benefits of sequencing the human genome:

- 1. Astounding commercial success:** The HGP has fetched around \$796 billion to the U.S. economy between 1985 and 2010, offering a staggering 3.8 million worth job years in the process. In 2010 alone, the genomics industry generated \$3.7 billion as nationwide taxes.
- 2. Disease prevention and diagnosis:** Predicting the onset, improve the diagnoses and discover ways to prevent incurable diseases like cancer, tumors, sudden heart attacks, [pregnancy high blood pressure](#), disability, etc.
- 3. Intelligent drug modification:** Whole human genome sequencing is also helping scientists trace and design effective drugs by evaluating the patient's genes to discover variants. In other words, genome study can enhance the efficacy of drugs which are otherwise ineffective for certain patients due to their intrinsic genes.
- 4. Application:** Whole Human Genome Sequencing has widespread potential implications on Energy industry, Molecular medicine, Forensics, Risk management, Breeding, Anthropology and Archaeology, among others.

Cons

1. Scope for financial exploitation: The sheer commercial parlance of genome sequencing leaves a lot of financial manipulation by pharmaceutical, federal and scientific bodies that are solely dictated by commercial motives. This is especially true in the case of customized drug manufacturing which can cause ambivalence to many.

2. Controversial: HGP is inherently a politically divisive topic that polarizes various groups/parties. Religious and other related organizations argue that whole human genome sequencing is a sophisticated strategy to tamper and interfere with the laws of the Divine and also nature. However, other progressive entities and scientists feel otherwise.

3. Loss of privacy: HGP also runs the risk of government agencies and other agencies using your genetics information against you by 1) Sharing it with other organizations for research purposes; 2) denying insurance on the basis that your genes make you liable to suffer from grave diseases; and 3) Dictate medical policies using unfair practices.

<http://www.healthfitnessinfomagazine.com/biotech/think-you-know-human-genome-sequencing-pros-and-cons-think-again/>

As Genome Sequencing Becomes more Affordable Should You Do It?

Genome sequencing is becoming more affordable than ever before – several companies in the industry say the [\\$1,000 personal genome](#) is just around the corner. But, even if you can afford it, is mapping your genes worth it if you don't have a specific medical condition to consider?

Despite the whole “knowledge is power” argument – it could help with early diagnosis and prevention or lead a doctor to better treatment options for an existing condition – sequencing skeptics raise valid concerns and questions when it comes to gene sequencing for healthy people. How precise is it? How well will consumers be able to interpret the results? Will it just lead to needless hand-wringing about conditions that people won't be able to do much to address or that won't surface until much later in life?

For now, those are questions for people with only the deepest pockets. But it won't be long before the conversation becomes more relevant for more of us and, in the [Wall Street Journal](#) this week, two doctors weigh in with the pros and cons of the debate over whether healthy people should have their genomes sequenced.

Dr. Atul J. Butte, division chief and associate professor at the Stanford University School of Medicine and director of the Center for Pediatric Bioinformatics at Lucile Packard Children's Hospital in Palo Alto, Calif., takes the pro position. And Dr. Robert Green, a medical geneticist at Brigham and Women's Hospital and Harvard Medical School in Boston, argues against it.

Even though Dr. Butte acknowledges that gene sequencing isn't perfect, he believes the positives outweigh the negatives. He says:

- Identifying DNA variants that are early indicators of disease can lead to early diagnoses and preventative strategies.

- Couples planning families can learn whether they carry genetic risks for serious disorders.
- Doctors can better figure out the most effective drugs for a patient or what to avoid
- It can help in the diagnosis of illnesses that haven't yet been identified.

On the flip side, Dr. Green believes that while affordable genomic analysis opens the door to personalized disease prevention and treatment options, there are still roadblocks. For example:

- Medically dangerous gene mutations are rare in healthy individuals but it would still be very expensive to find them – less than 2 percent of healthy people have a dangerous DNA mutation that would spur a doctor to monitoring or treatment. Assuming sequencing costs \$5,000 now, it could cost \$250,000 to find one person with a mutation.
- Known mutations may or may not carry the same risk without a family history, so sequencing alone can't always lead to action.
- Geneticists don't always agree on whether gene mutations are dangerous.

When it comes to health, I tend to fall on the side of information – the more of it we have, the better off we are. And the rise of consumer-ready medical technology that gives us clearer windows into our bodies – from [Fitbits](#) (see disclosure) to the [AliveCor](#) iPhone-compatible [heart monitor](#) – is setting the stage for an era in which people are armed with even more data about their health. [23andme](#) doesn't do full gene sequencing but its genotyping services already let people explore their DNA for just \$99.

But as we move into this new bioinformation-filled future, it's important to keep the skeptic's voices in mind because gene sequencing doesn't just have personal implications but public health consequences. One of Dr. Green's most haunting concerns is the rise of "patients in waiting" who spend their lives in anxiety, undergoing unnecessary tests and potentially doing themselves more harm than good. But as [others have noted](#), sequencing could take its toll on the health care system with unessential screenings and procedure, tax the patient-doctor relationship and lead to [other biotethical questions](#).

Genetic Health Pros Cons

Genetic testing may be beneficial in determining whether or not you have a disease or are likely to develop a disease over the course of your life. By examining your DNA, geneticists can look at variations in DNA sequences called genetic markers that indicate a person's predisposition to developing an inherited disease that may run in their family. Geneticists may be able to see how likely you are to develop cancer years before the disease takes over your body.

Pros and Cons of Genetic Testing

The pros and cons of genetic testing depend on how a person will benefit from the test results. If a genetic test reveals a strongly predictive negative result, a person probably will not develop a disease and frequent doctor visits and medical testing can be eliminated along with endless worry and uncertainty. In other words, if a test reveals the desired result, the advantages are relief, and there really are no disadvantages.

On the other hand, if a gene test demonstrates a genetic predisposition to an inherited disorder, the news can be depressing and a patient can be burdened with information over which they have no control. However, knowledge of predisposition may give the person enough time to take preventative measures, including taking medicine or adapting lifestyle changes to lower the risk of contracting a disease.

If a disease is caught early on, the patient has the best possible chance of survival. Understanding the advantages and disadvantages for genetic testing are important in deciding whether or not to undergo a genetic test.

Disadvantages of Genetic Testing

DNA genetic testing can be quite expensive and may not be included under a patient's insurance policy. Some genetic tests may require additional testing, which you may or may not be able to afford. For example, a genetic test from an online DNA testing company may cost between \$620 and \$3,456. There is the possibility that a person could spend thousands of dollars and not be able to benefit from the results.

It is important to understand that there are no real certainties in genetic testing. Genetic testing only indicates probabilities, which are not 100 percent certain. Despite breakthrough advances in DNA

<http://morefocus.com/health/genetic-health-pros-cons/>

genetic testing, gene tests simply cannot rule out every possibility of developing a disease. Positive tests also do not necessarily mean a patient will necessarily develop a disease or disorder soon, if ever. Thus, there are certain limitations on scientific research technology and the margin of error of certain tests.

If a genetic test reveals you are at risk for a genetic disorder, there is the possibility that other family members may be at risk. If a genetic test reveals you are at risk for developing an inherited disease, whether or not you decide to share your status with family members is both a personal and ethical choice you will have to make.

Genetic Testing, Ethics and Concerns

Ethical issues are a definite concern and one of the disadvantages for DNA genetic testing. Issues in genetic testing ethics center around privacy and who should have access to the results of DNA genetic testing. Could a person be denied insurance coverage if he or she is likely to develop cancer later in life? Because there are not cures to every inherited disease, there is potential that the results of a genetic test could do more harm than good. Sharing your health risk status could potentially lead to discrimination at your job or for your medical coverage, no matter how illegal such discrimination might be.

<http://morefocus.com/health/genetic-health-pros-cons/>
Whole Genome Sequencing

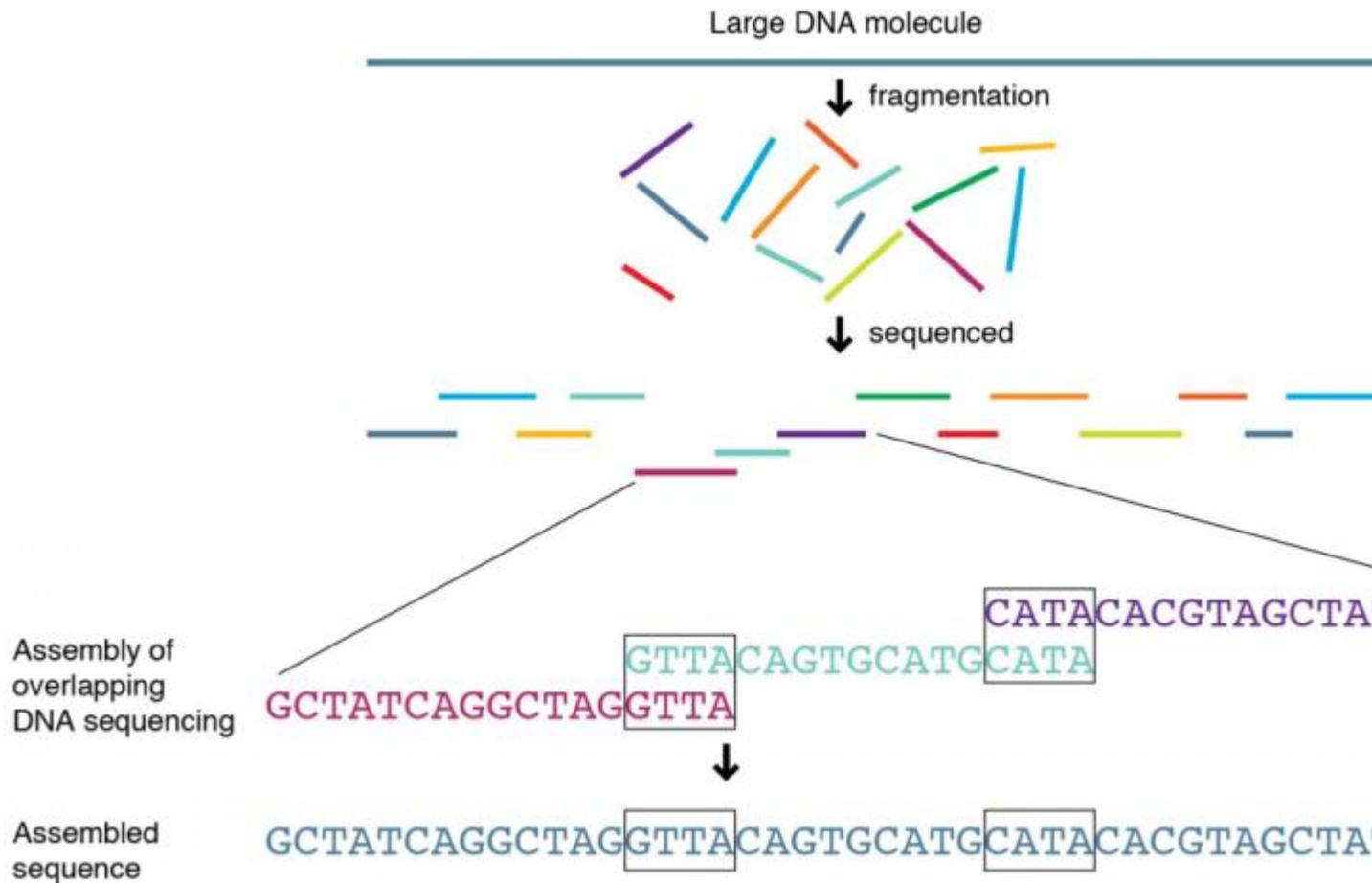


Image Courtesy of National Human Genome Research Institute

During whole **genome** sequencing, researchers collect a DNA sample and then determine the identity of the 3 billion nucleotides that compose the human genome. The very first human genome was completed in 2003 as part of the **Human Genome Project**, which was formally started in 1990. Today, sequencing technology is much more efficient, and a human genome can be sequenced in a matter of days for under \$10,000. The first human genome cost \$2.7 billion. Today, most genetic testing focuses on one or a few genes, rather than the entire genome. However, with the falling cost of genome sequencing, more individuals are pursuing this option. Physicians can look at an entire genome to see how specific treatments for a disease will be affected by an individual's unique genetics. For example, the physician may opt to look at genes involved in drug metabolism when deciding dosage. In the future, whole genome sequencing may enable everyone to develop a personalized treatment plan.

Advantages of Whole Genome Sequencing

- Creating personalized plans to treat disease may be possible based not only on the mutant genes causing a disease, but also other genes in the patient's genome.
- * Genotyping cancer cells and understanding what genes are misregulated allows physicians to select the best chemotherapy and potentially expose the patient to less toxic treatment since the therapy is tailored.
- * Previously unknown genes may be identified as contributing to a disease state. Traditional genetic testing looks only at the common "troublemaker" genes.
- * Lifestyle or environmental changes that can mediate the effects of genetic predisposition may be identified and then moderated.

Disadvantages of Whole Genome Sequencing

* The role of most of the genes in the human genome is still unknown or incompletely understood. Therefore, a lot of the "information" found in a human genome sequence is unusable at present.

* Most physicians are not trained in how to interpret genomic data.

* An individual's genome may contain information that they DON'T want to know. For example, a patient has genome sequencing performed to determine the most effective treatment plan for high cholesterol. In the process, researchers discover an unrelated allele that assures a terminal disease with no effective treatment.

* The volume of information contained in a genome sequence is vast. Policies and security measures to maintain

the privacy and safety of this information are still new.

The Pros and Cons of Genetic Testing

<http://www.nextavenue.org/article/2013-04/pros-and-cons-genetic-testing>

A \$99 at-home test can detail our genome profile and assess our risk of contracting disease. But is that good news or bad news?

By Rita Rubin | April 12, 2013



iStockPhoto/ThinkStock

The era of personalized medicine is coming. As genetic profiling, also known as [genome sequencing](#), gets better, cheaper and more widely available, more of us may one day know exactly which chronic diseases we're likely to contract, what medications we should avoid and even what type of diet we should follow. In just a few years, with such data in hand, our doctors might be able to identify our vulnerabilities and target treatments with a greater chance of success than ever before.

We can get a preview of that future today through private services like [23andme](#), which, for just \$99, will analyze a half-teaspoon of your saliva and return a detailed report with some 250 bits of information about your genetic profile and how it could influence your health.

It's far from a full genomic sequencing, but it does provide some potentially valuable data. It's also something of a bargain. When the company launched its service in 2007 — the name is inspired by the 23 pairs of chromosomes that make up the human genome — the analysis was priced at \$999 and provided far less information. The clinical cost of complete genome sequencing is more than \$3,000, though it is also falling.

"For 99 bucks you have a pretty good set of information," says Dr. Eric Topol, a cardiologist who is the chief academic officer of Scripps Health in San Diego and the author of [The Creative Destruction of Medicine: How the Digital Revolution Will Create Better Health Care](#). "But it's only a partial story."

One Woman's Genome

Misha Angrist, an assistant professor at the Duke Institute for Genome Sciences and Policy and the author of [Here Is a Human Being: At the Dawn of Personal Genomics](#), was one of the first people to view the results of the sequencing of their entire genome, and is one of many who now wonder how popular direct-to-consumer genetic testing will become. "In general, people who give up their credit card numbers and their saliva are a self-selected bunch who are interested in this information," he says. (Testing is available to consumers nationwide, with some exceptions: Maryland bars the shipping of genetic tests to consumers, and New York forbids shipping your sample from within the state.)

I snapped up a 23andMe test kit in December 2011, becoming one of more than 200,000 people so far who has enlisted the service. I was concerned about whether I carried a genetic mutation that would greatly increase my risk of breast and ovarian cancer — my father's mother and one of his sisters had breast cancer. While only about 10 percent of all cases are considered hereditary, scientists have been able to link three genetic mutations to virtually all inherited breast cancers in Ashkenazi (Eastern European-descended) Jews, like me.

Though I rushed to order the kit, I confess to waiting several months before sending my sample to the company's California lab. I was nervous about what I'd find out. After I got the notification that my results were available, I waited until my next doctor's appointment to click on the link that would tell me whether I had one of the Ashkenazi mutations. Much to my relief I was negative for all three, but I'll continue to get mammograms and clinical breast exams because the 23andme assessment doesn't ensure I'll never develop breast cancer.

What You Learn

The information from services like 23andme is not complete. Generally, it can tell you only if your odds of a certain condition are better or worse than the general population. Testing can reveal, for example, if you have a variation in your genes that increases your odds of developing diabetes from one in five — the general population's risk — to one in three. If you discover that you have that variation, you may decide to act on the news by changing your exercise and diet regimens to ward off the condition.

Testing can only reveal what scientists know so far about the link between genes and disease. It can tell you if you have the rare genetic mutation that appears to raise one's lifetime risk of contracting Parkinson's disease to 60 percent, but not having the mutation doesn't at all mean you're in the clear. My own results indicated that my risk of contracting Crohn's disease, an inflammatory bowel condition, was only about half that of the general population — but I've already lived with the disease for more than 20 years.

In the case of heart disease, testing can show you if your ethnicity, or the presence of certain genetic markers, indicate an elevated risk. As is the case with many conditions, though, environment and lifestyle play at least an equally important role in determining your actual risk of heart disease. The company's reports detail which conditions are more or less affected by genetics. There are certain concerns for which research indicates that genetic tendencies play a larger-than-average role in determining one's risk. 23andme estimates, for example, that genetic factors determine at least 60 percent of an individual's risk of developing obesity.

The Best Value in Testing

Perhaps the most reliable, and immediately actionable, component of the 23andMe data is its assessment of how we will respond to certain drugs. For example, the company checks for genetic variations related to how well we metabolize clopidogrel (Plavix), which helps prevent blood clots that could cause heart attack or stroke, and warfarin (Coumadin), a blood-thinner that prevents and treats clots in the veins, lungs and heart.

Clopidogrel needs to be converted into its active form in the liver. Most of us have two functioning copies of the gene that carries the blueprints for the enzyme needed to activate the drug. But some of us have only one, and about 1 in 20 people have none. Those in the latter group are considered to be poor metabolizers of clopidogrel; for them, the drug works about as well as a sugar pill.

About a third of patients prescribed warfarin turn out to be more sensitive to the drug than expected, due in part to genetic mutations. This is why, in 2007, the Food and Drug Administration approved labeling on the drug that suggests doctors use genetic tests to optimize dosing. For people with a heightened sensitivity to warfarin's effects, a dose of the drug that is too high puts them at risk of potentially life-threatening internal bleeding. If a dose is too low, though, potentially lethal blood clots could form.

The Psychological Element

I'm not sure how I would have reacted if I had learned I had a genetic mutation increasing my risk of breast cancer. Experts remain concerned about the psychological impact of finding out that you're at high risk of a potentially fatal disease, especially when companies can send you the information via email without the context and advice your doctor could provide in person.

There's a threat to the health care system, some clinicians believe, in producing a legion of "patients in waiting" — otherwise healthy, asymptomatic people who have found out they may be at risk for chronic disease, then overload hospitals with demands for further testing and treatments that may not be necessary.

(MORE: [New Prescription for Better Health Care: Less Is More](#))

Several studies are under way to find out just what actions people — and their doctors — take based on genetic data. Topol says his own research has shown that patients don't necessarily panic when they learn the results of direct-to-consumer genetic testing. (23andMe advises that its information "is intended for research and educational purposes only, and is not for diagnostic use.")

For patients with a risk of disease but no symptoms, the wisest action would be preventive steps, like changes in diet and lifestyle to ward off heart disease. But even with genetic data in hand, many of us may not take steps to improve our health. "I'm fairly comfortable with the conclusion that people don't freak out," Angrist says, "but neither are they likely to change their behavior."

Millions of us resist giving up cigarettes or fatty foods, despite the obvious health risks. When we find out that our genes make us

even more susceptible to chronic disease than we thought, will we respond differently? That's one of the things we'll discover as genome sequencing goes mainstream.

<http://www.nextavenue.org/article/2013-04/pros-and-cons-genetic-testing>