The Human Genome Project at 10 years: A teachable moment

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It's now been 10 years since humans deciphered the digital code that, in a very real sense, defines us as a species. It's hard to overestimate the significance of that achievement but easy to misconstrue what it means and where its true promise lies.

The philosophical import of having the human genetic code in hand is remarkable and we've learned much that is fascinating. For example, we scientists were surprised (and humbled) to discover that it takes only about 22,000 genes to build a human being—half the number it takes for corn to get on with its business. The Human Genome Project, which came in ahead of schedule and under-budget (at about $3 billion), has also shed light on some very big questions about human ancestry and evolutionary biology.

But the project wasn't sold to the American people to answer evolutionary questions, ferret out how humans populated the globe, or discover interesting factoids, no matter how fascinating or important these subjects might be. No, what the American people want from their investment is better medical care, and it was this promise that fueled the project's funding and its popularity.

Now at the 10-year mark, we are witnessing arguments about whether mapping the human genome was worth the money and effort. People are asking where all that genomic medicine is that we were promised a decade ago. So it's a good time to take stock of where current and future genomic payoffs might reside and where we may have fallen short. But it's also a rare opportunity to illuminate more fundamental issues related to the nature of scientific progress, how the public and scientists communicate, and what we really should expect from science.

The Human Genome Project has certainly yielded some practical benefits to clinical medicine. One of the near-term successes is the field of pharmacogenomics, in which knowledge of a patient's genomic information is used to guide drug choice and dosing. Specific agents—such as abacavir for the treatment of HIV, and clopidogrel, a drug widely used to reduce the risk of heart attack—can be more precisely prescribed now that we understand how individuals' genomic constitutions influence their responses to these agents. Pharmacogenomic approaches won't be applicable to every drug but for select agents, knowledge derived from the Human Genome Project is already improving the care of patients. In another near-term advance, our ability to better diagnose rare genetic diseases (which are no less tragic to those who have them simply because they are rare) is gaining tremendous traction because of knowledge and technology propelled by the project.

We've gained insight into the molecular underpinnings of many common diseases and found variants in the population that are related to an individual's risk of almost every disease imaginable. This knowledge will provide long-term benefit through new drug targets and better understanding of those diseases. But any direct or practical payoff from such knowledge is years away. The idea that you will benefit in any tangible manner from knowing whether you are at an increased or decreased risk of say, heart disease, is a fool's hope despite early delusions by scientists and a fledgling direct-to-consumer genetic testing industry that would have you believe otherwise. The simple fact remains that whether you are at double or half the population's average risk for a given common disease isn't very important. You are still highly likely to develop them. That's why they're called common diseases. Moreover, we've known for a long time how to prevent such diseases: eat right, drink responsibly, exercise, and don't smoke (i.e., your mom was right). As could have been predicted by any practicing physician 20 years ago, parsing an individual's risk for most such diseases adds little to information easily gained by taking a brief family history, asking a few questions, and ushering you onto a scale.

As in all scientific pursuit, the true promise of deciphering the human genome lies in the gradual and incremental accrual of basic knowledge. Ultimately, the better we understand ourselves and our world, the better we will be able to act on that knowledge and improve our lives. Because of scientific progress, your life is unimaginably richer and more comfortable than that of any human who ever lived, even just a few generations ago. But the timeline is agonizingly long for the translation of scientific knowledge into practical benefit. The road to tangibly better lives from scientific insights is unpredictable and filled with dead ends and U-turns. That's the nature of science and there are no shortcuts.

Both scientists and the public must come to terms with the true nature of science, or we will continuously have the same pointless debate about whether developments and discoveries live up to their promise. Scientists need to resist the temptation to oversell the short-term benefits likely to be derived from basic insights and advances. This is especially important in the realm of medical care, where the stakes are enormous, the variables are many, and our chances of being misled are high.

Inflating an unrealistic bubble of expectations not only ignores the reality of how science works but also it is bad politics on the part of scientists. For when the bubble bursts and the cool things we've been promised don't materialize (hey, it's 2010, where the hell is my hover car, anyway?) legitimate scientific pursuit suffers to the detriment of all. But it's not all the scientists' fault. The media must resist feeding the bubble of unrealistic expectations in search of the next good story. Finally, the public has a responsibility to understand the nature of science, to shed simplistic expectations, and to fund quality basic research with their tax dollars without expecting immediate benefits.

Just as scientific knowledge has transformed our lives (mostly) for the better over and over again, the Human Genome Project too will transform medicine and was well worth the cost. Its payoffs will be pervasive but will be predictably incremental—evolutionary rather than revolutionary.

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