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## The True Cost of Whole Genome Sequencing



By Dr. Robert C. Green

A few years back, you might have heard people talking about “the thousand-dollar genome”—the idea that sometime in the near future, you would be able to have your entire genome sequenced and analyzed for less than \$1,000. The idea was that once technological improvements lowered the price of sequencing to this price point or even less, we would have reached a tipping point that ushered in a new age of genomics with incredible implications for human health.

That future has now arrived, in that at least one company is selling consumer directed, physician-ordered whole genomes for under \$1,000, and there is an expectation that prices will continue to fall. Indeed, last year, the (then) new CEO of Illumina made headlines by predicting that “within a few years” the price of at least the technical component of sequencing would drop as low as \$100!

But the true costs of genome sequencing don’t end with the technical production of A’s, T’s, C’s and G’s, and they don’t even end with the molecular interpretation of a few dozen or even a few hundred genes. Central among these concerns: What happens after the genome sequencing results come back? Will patients end up paying more for potentially unnecessary follow-up testing than they did for the sequencing itself? Will clinicians spend an inordinate amount of time dealing with all of this, enough to put a strain on healthcare systems and raise costs for everyone?

Concerns about unnecessary testing and downstream costs are not just hypothetical. In a study published last year, researchers tracked more than three million patients in Ontario who had received a routine annual health exam and whose medical records showed no serious risk

factors for cardiac disease. Despite clinical guidelines that caution against routine electrocardiograms (ECG) for low-risk patients, the researchers found that 21.5 percent of patients received an ECG within 30 days of their routine health exam. Patients who went back for an ECG were five times more likely to have further cardiac testing or consultation. As the researchers put it, routine ECG testing increased the likelihood of a “cascade” of further testing and evaluation—even though the rate of actual cardiac problems was very low for both groups, whether or not they received a follow-up ECG.

The worry is that the same thing could happen with whole genome sequencing. If it becomes routine for apparently healthy patients to have their genome sequenced, will this lead to a cascade of costly and unnecessary follow-up?

Over the past few years, in the MedSeq Project, we have attempted to interrogate *every* gene associated with a well-established disease, seeking to define and communicate to patients and their providers any valid monogenic risk. Because we looked at so many genes, we discovered a startling 15% of our participants received unanticipated monogenic risk findings. And because we conducted this experiment as a randomized, controlled trial in which research volunteers were assigned by chance to be sequenced or not, we were in a strong position to compare the two groups, and to rigorously examine the benefits, risks and downstream costs associated with sequencing and disclosing genomic information among people willing to learn this information.

Our analysis of the “econogenomics” of participants in the MedSeq Project was led by Kurt Christensen and recently published in the journal *Genetics in Medicine*, and our findings differed from the ECG study’s in a few crucial ways. In the MedSeq Project, half of each group received, through their medical providers, whole genome sequencing along with a family history report, while half received only the family history report. We tracked participants for six months after receiving their reports and estimated downstream costs.

To be clear, the whole genome sequencing itself was not cheap. The cost of the actual sequencing, interpretation and report preparation was around \$5,000 per participant. But the most interesting findings from this study compared the average downstream costs between those who received whole genome sequencing and those who did not. Over the six months following the disclosure phase of the study, there were slightly more healthcare visits scheduled by those who received results of WGS, but the differences between the two arms (sequenced vs non-sequenced) averaged less than \$1,000 and was not statistically significant. This equivalence remained whether or not the analysis included hospital costs.

If these results hold in other scenarios, then the cost of implementing whole genome sequencing throughout the practice of medicine will be driven mostly by the costs of sequencing and interpretation, not the costs of medical follow-up. Since sequencing is generally accepted to

be something that will eventually become ubiquitous and will help move clinical medicine toward personalized and preventative care, this is good news.

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