

# Econogenomics: The Economics of Genomic Testing for Health



Genomes2People

Jan 29 · 4 min read

By Dr. Robert C. Green



Let's say it was possible to buy your health by the day. How much would you be willing to pay for each year of perfect health? What if you could buy years of health for your loved ones, too? At what price point would you draw the line?

This sort of difficult calculus, on a much larger and chronologically longer scale, underpins many decisions we make in medicine — not just decisions that we make as patients, but also the decisions that are made for us by employers, health insurance funders and policymakers. We don't have the resources to pursue every possible treatment, to research every possible breakthrough, so how do we allocate the resources available? It turns out that there is an entire field of healthcare economics devoted to

understanding the costs and benefits of conventional medicine, and to navigating the trade-offs between more expense and better healthcare.

Determining the costs and benefits of new areas like genomic medicine is especially tricky, because we have so much less experience in these areas, and even experts cannot yet fully agree on the spectrum of harms and benefits.

Kurt Christensen, PhD, Instructor in Population Medicine at Harvard Pilgrim Healthcare, is a leading researcher working on these questions, along with an Econogenomics Working Group we established in 2018. The group recently published two papers that built on data collected in the MedSeq Project, the first randomized trial of comprehensive sequencing and interpretation in apparently healthy adults. One paper analyzes short-term costs of whole genome sequencing (WGS) in medical settings; the other essentially outlines why that is so hard to do.

“A lot of what we’re pioneering is simply how to do these studies, how to measure cost-effectiveness of genetic screening at a population level,” Christensen said. He explained that while most existing work has looked at costs and benefits of interventions for people already diagnosed with a disease, it is more difficult to quantify benefits of genomic screening for apparently healthy people.

The first of these econogenomics studies followed people who received WGS results, including apparently healthy people who received their results through a primary care provider. We estimated the costs of sequencing and the downstream costs over a span of six months, finding some reasons for optimism about the long-term costs of WGS.

But the information one receives from WGS can have implications far beyond six months, potentially influencing medical and reproductive decisions for the rest of someone’s life, and their sequencing data can be re-analyzed periodically to uncover gene-disease associations we didn’t previously know about. This is a lot more difficult to study, for many reasons.

For one, Christensen said: “How do you fund a study that’s going to follow someone their entire lifetime?” Consider the people participating in the study, too. “Even following a family for a year in a randomized controlled trial is a lot to ask of them.”

There is also the problem of timeliness. “Trials capture the real-world messiness of how patients and providers respond to genetic information,” Christensen said, but policymakers and others need to make decisions now — not in ten or twenty years — about how to handle these services, including whether to cover them.

Econogenomic modeling could address some of this. We’re working on approaches to use what we’ve learned from trials to perform projections about health outcomes of genomic medicine. One early takeaway: don’t get too hung up on the short-term costs.

“The cost of sequencing itself is already comparable to a lot of other diagnostic tests regularly used in medicine,” Christensen said. Besides: “What really moves the needle on cost effectiveness isn’t the costs — it’s the benefits.”

And these are often difficult to quantify. What’s the value of just providing the name for a condition, to help patients or parents understand what’s happening, even if no treatment is available? What happens if that allows that patient or that entire family to be first in line for a newly developed treatment that salvages their health or saves their life?

Clearly, there are many unknowns, and we have our work cut out for us, but if we are to integrate genomics into medicine with less than perfect knowledge of all the risks and benefits, we will have to take economic modeling of this nature seriously, and use it to make decisions about the implementation of genomic medicine and the reimbursement of preventive care.

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