



EDIT

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# Genomic Sequencing: Just Another Tool in the Doctor's Bag



By Robert C. Green, MD, MPH

Futurists are predicting that genomic sequencing will profoundly disrupt the practice of medicine! But from the stethoscope to the x-ray, medicine has successfully integrated new technologies before. Will this be different?

This is one of the questions we have been asking in the [MedSeq Project](#), the first randomized clinical trial funded by the National Institutes of Health to explore the integration of sequencing into “everyday” medical practice. In the MedSeq Project, we are investigating whether primary care doctors can integrate findings from genome sequencing of healthy individuals into their practices, and asking what both the doctors and their patients will do with this information.

Early findings from the MedSeq Project suggest that “disrupting” medicine may be overstating the case. Our early data suggest that physicians involved in daily patient care can learn to use genomic information appropriately, and that genomic findings simply provide more data for physicians to gather and process about their patients.

Whole genome sequencing (WGS) and whole exome sequencing (WES) are still new to the practice of medicine, used mainly at this time for care of patients with cancer and to discover molecular diagnoses in patients with mysterious clinical presentations. The argument is often made that physicians outside of oncology are woefully underprepared to use these technologies in the day-to-day practice of medicine, and may resist its application. Even trained geneticists are not entirely comfortable with the use of sequencing in daily practice.

Furthermore, there are fewer than 2,000 board-certified medical geneticists and just over 4,000 genetic counselors nationwide — far too few to meet the anticipated escalation of genomics in medicine.

First, in the MedSeq Project, the doctors we are studying expect to incorporate genomic information into their practices over time. “Someday it will be the norm,” one of our study subjects told an investigator. A [recent MedSeq publication](#) supports how ubiquitous this sentiment is among at least some types of medical practitioners.

But how will doctors get from where they are now (unfamiliar with genomic technologies and uncertain about how to use them) to where we all agree they need to be? Dr. Jason L. Vassy, Dr. Bruce R. Korf and I took a look at this question in a [study published earlier this year](#). We began by noting that doctors may feel “unprepared” for genomic information in the same way they were unfamiliar with CT scans in the 1970s, but we reminded readers that today’s doctors are comfortable ordering CT scans for their patients, and find the resulting information invaluable in patient treatment. We also highlighted new continuing medical education (CME) programs and [other resources](#) developed by the National Human Genome Research Institute that will facilitate the process.

Just as doctors don’t have to be domain specialists in chemistry to use chemistry lab tests, or in radiation science to use x-ray reports, we imagine that the physicians in the near future will

quickly become skilled at using genomic information that is delivered in an understandable format by a molecular laboratory and will be able to contextualize this information to a patient's family history, medical history and physical examination. Better electronic health records and decision support tools will help this process, but doctors will not wait for the complete development of seamless systems if genomic information proves to be medically useful.

In order for this to happen, there will need to be a short and understandable "genome report" that doctors can use (just as today they use an x-ray report). In the MedSeq Project, we have published [descriptions of the variant classification strategy we have used](#), and an early imagining of what that format might look like. In fact, we have created a [one-page whole genome report](#), and this is exactly the intervention we are testing in our research.

The MedSeq Project is also asking what doctors will do with sequencing information once they have it. As described in the MedSeq Project methods paper, the focus of the randomized trial has been to engage primary care physicians and cardiologists in receiving clinically relevant WGS and interpretation for some of their patients and to observe how this information is incorporated into clinical care. None of the doctors were card-carrying geneticists and no genetic counselors were involved in the direct care of the patients, but the doctors in the study went through a brief educational orientation about genomic information that reviewed inherited diseases through case studies.

Since the doctors received their patients' genomic sequences, we are finding that some of them use this new information to alter treatment plans. Our researchers are interviewing these physicians about whether they found genome sequencing useful, and how its usefulness compared to traditional family histories. In early analyses, a few trends are already clear.

The doctors in our study told us that they still consider family

history more useful than genome sequencing. They believe that family history will remain a valuable clinical tool, but they do expect WGS to be increasingly useful, even inevitable.

One doctor expressed a common view on a current challenge to genome-informed medicine: “I think for a long time there will be too many genes and not enough correlation to disease, and that ratio will have to gradually change so that we understand more about the specifics.”

As research continues, the use of genomics in day-to-day medicine seems almost certain. “I don’t know when, but I’m sure sometime in the future it will become very much a part of what primary care physicians do,” another of the doctors in our study said. “It will be just one of the tools in their toolbox that’s available for taking care of patients.”

The MedSeq Project imagines a near-term future in which genomic information is not the realm of specialists, but is one tool among many in all doctors’ clinical practice. Our early findings show that some doctors in private practice share that vision — although to be sure, these doctors aren’t representative of our national population of physicians. The doctors within the MedSeq Project are self-selected early adopters in an academic medical center. Yet, while most of them have little background in genetics, they are not only managing the genomic data with their patients, but are using WGS results to complement other clinical information, identify risk categories, motivate patient behavior, and customize medications to genomic profiles.

As more individuals seek out information about their own genomes, physicians will increasingly find themselves using genome sequencing and other advanced technologies as one more way of personalizing patient care. There simply won’t be enough genetics specialists to manage all of the medically relevant information when this scales to millions of individuals... we will have to roll up our sleeves and find ways to make this happen responsibly.

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