How Genomics Could Save Your Life

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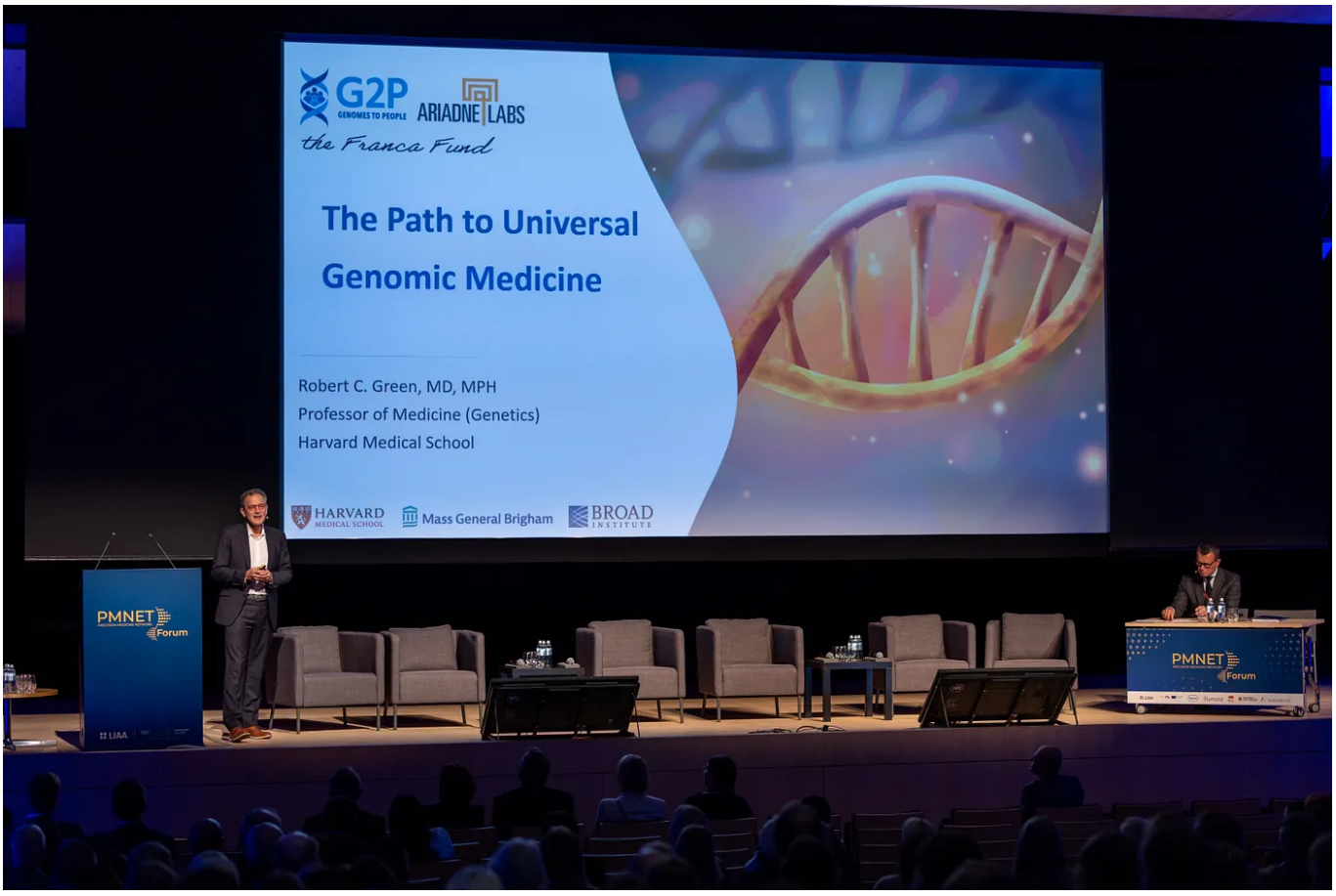
What's holding us back from doing more with preventive genomics in medicine?



By Robert C. Green

Adapted from my keynote at the Precision Medicine Network (PMNET) Forum in Riga, Latvia.

I'd like more people to ask themselves: Why is the healthcare system not using genomics in my everyday healthcare?

We could all of us, right now, understand what's in our genome. For some of us, it could be lifesaving.

Precision medicine covers everything from smart watches to liquid biopsies, but genomics is the tip of the spear. Near-term applications are already here: diagnosis of rare conditions, some limited pre-conception carrier screening and embryo selection for hopeful parents, and various uses in cancer diagnosis and treatment. But I am most excited about the next phase of potential for genetic testing: newborn and childhood sequencing, expanded carrier screening for expecting parents, polygenic risk scores, new pharmacogenomics applications, and population screening for all age groups.

So why is preventive genomics not already used for these things?

There are reasonable concerns about privacy and discrimination if genetic information is misused or falls into the wrong hands, and worries that people (both patients and healthcare providers) won't understand the information and will take harmful action as a result. We worry, in short, that preventive genomics could cause more harm than benefit. Much of our own research has asked: What are the medical, behavioral, and economic outcomes associated with unanticipated findings from genomic sequencing?

We studied this over a decade ago with MedSeq, the first randomized trial of comprehensive genome sequencing in apparently healthy people. In this study, we gave genomic sequencing data to participants through their primary care physicians and followed up to see how they — and their doctors, who we trained to read the results — responded to this information about themselves. In the process, we made a startling discovery. We found that 20% of apparently healthy adults were carrying a monogenic disease risk. This doesn't mean you *have* the disease, but it does mean that you have a significant risk of expressing the disease at some point in your life.

Even more interestingly: Those 20% of participants reported no significant difference in anxiety after receiving their results compared to the other 80% of participants. The doctors weren't overwhelmed with the genomic reports, and they relayed accurate information to the patients with very few errors. We even tracked downstream medical referrals and follow-up testing and found that participants' healthcare spending didn't go through the roof in response. In short, the benefits outweighed the harms.

With BabySeq, we did something similar with newborn and infant screening, vastly expanding the standard screening experience with whole exome sequencing and interpretation of approximately 1,000 genes. We found unanticipated and potentially important mutations in 11% of newborns who had not previously been suspected of having genetic diseases. These results allowed parents and providers to look more closely for clinical indications of those specific conditions, and in many cases to take preventative actions that could have enormous benefit for the rest of the child's life. And once again, we found that a few months later, parents of those 11% of newborns with clinically important mutations weren't any more anxious or distressed than the other 89% of parents in the study.

There's another way we can save lives in the very near term: We can return incidental genomic results to research participants who undergo any form of genetic testing. About 100 million people in the world have been sequenced for research, and across the surveys of those participants, 70–80% said that they wanted their genetic results returned to them. Yet this is almost never done, due to a combination of concerns about logistics and participants' "right not to know" — and perhaps because it is more work and more expense for the researchers, requiring thoughtful design at the front end of the study. Indeed, in 2021, we coordinated a recommendation paper from the Global Alliance for Genomic Health that recommended strong consideration of returning incidental or secondary findings in every research project that utilizes genetic testing!

Again and again, people tell us that they want this information. One BabySeq parent told us, after learning through the study that her son had a genetic heart condition: "I feel more empowered to have the information . . . If we know more, we can do the best for our kids." Another parent said: "Knowing any additional information is going to be helpful down the road." And across the board, parents said that if given the opportunity, they would do it again. You can hear this directly from the powerful voices of parents who have screened their children through BabySeq.

I believe we should treat genomic information more like we treat other medical information, sharing it more often with individuals and trusting them, and their healthcare providers, to be able to handle it. We could be focusing on helping people to understand their genomic sequencing data, rather than shielding them from it.

If we're going to respect the autonomy of patients and participants, we need to ask them if this is something they want. If they do want it — and many, if

not most of them, will want it — then we need to be prepared to deliver it to them.

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