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Exponential Medicine Conference and the “Last Mile” Problem in Genomic Medicine



By Robert C. Green, MD, MPH

I was invited to speak at Singularity University’s amazing [Exponential Medicine](#) event this winter, and it was one of the most remarkable meetings I have ever attended. In each and every presentation, rigorous science mixed with imagination, entrepreneurship mixed with mission, and the enthusiasm and appetite for new ideas were palpable among the audience and speakers.

Many of the ideas involved collecting or channeling “big data” into discoveries or products, and in my field of genomic medicine, big data studies are certainly all the rage. Advances in genome sequencing and computing allow researchers to comb through ever more immense heaps of data in search of medically important genetic variants, opening the door to a world of discoveries.

Our [Genomes2People](#) research program has a complementary research focus. Our work is in “small data,” focusing on rigorously measuring clinical utility and how to translate new advances in genomic medicine into the best outcomes for patients and families. By navigating this “last mile” of medical genomics, we can help understand how to change the marvelous discoveries of big data studies into real benefits for real people. If you are interested, you can watch the full video of the talk [here](#).

In the video, you'll see how we are creating rigorous small-scale studies of some of the first humans on earth to have genomics integrated with their everyday care. In the [MedSeq Project](#), we envisioned, designed, and have just completed the first randomized controlled trial to use whole genome sequencing with primary care doctors who are practicing medicine on the front lines. To do this we had to create an accessible and understandable [one-page Genome Report](#) as well as rigorously codify our genetic [variant classification methods](#). Using lessons learned from the MedSeq Project, we have begun recruitment for the first randomized controlled trial to use genome sequencing in newborn infants, the [BabySeq Project](#).

We are only beginning to scratch the surface of understanding the relationship between the massive number of genetic variants and their association with diseases and wellness. As we collect more information and make it available to individuals and doctors in clear and useful ways, we believe genomic medicine will become less intimidating to all involved. Indeed, despite much rhetoric to the contrary, we think that [physicians are as "ready" for genomics](#) as they have been for any new technological tools, and that physicians will soon be incorporating genomic information into their practice.

Please check out our [website](#) to learn more. We're actively recruiting genetic counselors, post-doctoral fellows, and research assistants for G2P projects so if you are interested in this area, we hope you'll [contact us](#)!

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