

The declining cost of genetic sequencing is opening the door for precision medicine

We live in an era where customization is convenient and frequently offered. People are used to ordering their perfect cup of coffee, swapping out ingredients and adjusting ratios to get it just right. Economies of scale help to achieve these available options, but there is still a cost associated with moving from generic to customized products. While it would be ideal to have all the clothes you buy custom tailored, the cost can be prohibitive, leading most people to buy their clothes off the rack. This is an apt analogy for our current healthcare system. Providing a custom tailored medical treatment plans may be greatly desired, medical professionals are often limited to what is available “on the rack”; treatments that were designed with large populations in mind, not individuals. There is an effort underway to reinvent medicine, one where steps are taken to incorporate individual patient traits into medical care. This initiative is known as precision medicine.

Precision medicine plans to take standard medical record keeping, with family history and treatment records for individual patients, and incorporate three new variables. Enhanced details about the patient’s lifestyle, environmental elements impacting their health, and patient genetic information are the three elements. These will help focus treatment plans and provide a higher degree of granularity in available options. How would precision medicine differ from the care one regularly experiences now? By integrating these additional elements of information into patient records, physicians would be able to prescribe drugs that might be able to offer better results with fewer side effects. Precision medicine aims to reduce the number of “shotgun” treatments, and replace them with more targeted options. An individual’s genetics can be used as a categorization or filtering tool, finding groups of people that have improved outcomes and reduced adverse effects of one drug over another. This practice is known as pharmacogenomics, and it is just one of the tools that would be used in precision medicine.

One of the biggest factors in enabling precision medicine to be developed for clinical practice is the falling cost of genetic sequencing. The ability to sequence an individual’s DNA, or reconstructing the proper order of the nucleic acids in their unique genetic code, has been possible for over a decade now. With technological advances, the cost to sequence a genome (the entirety of an individual’s genetic material) has exponentially declined (Fig. 1).

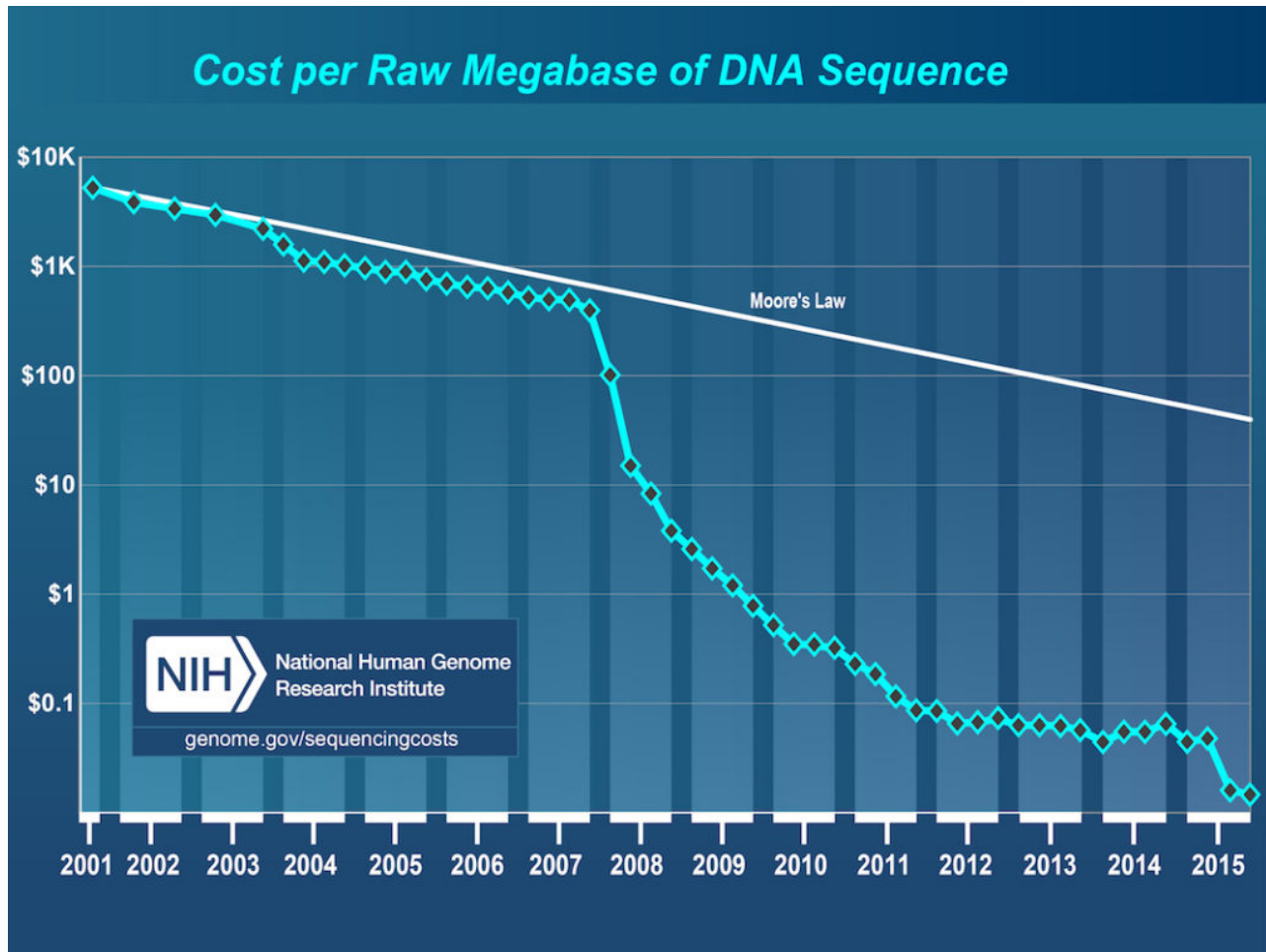


Fig. 1. Declining cost of genomic sequencing.

Wetterstrand KA. DNA Sequencing Costs: Data from the NHGRI Genome Sequencing Program (GSP) Available at: www.genome.gov/sequencingcostsdata. Accessed 9/20/16

What once cost tens of millions of dollars to perform for a single person is now close to dropping to under \$1,000. Targeted genetic sequencing approaches can be deployed for far less. Instead of mapping out the entire genome, tests can be run to search for specific errors, or traits with known increased risk factors. For example, searching for the *BRCA1* & *BRCA2* gene mutations, which is associated with greatly increased odds of developing ovarian or breast cancer. This decreased cost has increased the amount of genomic data researchers can explore to find new associations and enhancing our medical knowledge. It provides a foundation for the genomic component for precision medicine. There are several major obstacles facing the implementation of precision medicine. In our work, we outline several of these roadblocks, like current physician preparedness to address patient's questions about genetic testing results, and present some potential solutions. Precision medicine is an exciting evolution of medical care, however, detailed planning and

execution is necessary to reap the benefits and avoid doubling back to fix the mistakes made in haste.

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Publication

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