

Viewpoint: There's danger in overselling the benefits of routine DNA sequencing

For decades the potential of stem cells to cure all disease was promised. Today's reality is that the few worthy dividends reflect a very small part of the mostly unregulated landscape of stem cell profit centers overpromising, and often dangerously under-delivering. Now branded as Regenerative Medicine, the Food and Drug Administration (FDA) is forced to overcorrect the harms done by rogue stem cell enterprises in concert with a multi-country global effort by scientists to do the same (see [here](#) and [here](#), complications range from blindness to death). And the cycle repeats. Just substitute personalized or precision medicine for stem cells, and the fallacy of controlling your future by preventing the development of medical conditions draws a captive audience of willing participants, investors, and media attention.

The art of the sell

Consider the [latest pitch](#) from [Pennsylvania-based Geisinger Health System](#) as presented by its spokesperson Wendy Wilson,

“Geisinger is prescribing DNA sequencing to patients and putting DNA results in electronic health records and actually creating an action plan...We are preventing disease from happening.”

The conglomerate is building on an existing research biobank, offering DNA sequencing to 1,000 patients as part of their primary care visit contending it should be included like any other routine testing. Inevitably, their goal is to collect this data from their 3,000,000 patients.

[Geisinger](#) sets itself apart from more controversial direct-to-consumer genetic testing by maintaining it is mapping a focused portion of the genome called the “exome,” a functional area known to contribute to disease. The presumption is that their findings will result in medically actionable data as opposed to the wide net of genomic results reported by commercial products.

As CEO and President of Geisinger Dr. David Feinberg [writes](#),

“[Most of the medical spending](#) in America is done after people have gotten sick...We think this will decrease spending on a lot of care... [The way we look at it](#), that's millions of Geisinger family members who no longer have to rely on the law of averages to forecast their health and make plans about their life and how they live it...For these patients, precision health care reduces uncertainty and allows them to take charge. For clinicians, the technologies help us achieve, finally, what should be the true goal of medicine — keeping our patients well.”

Of course, as a health system, they offer, by patient request, a visit with a physician and genetic counselor for interpretation.

genome

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Let's unpack the smoke and mirrors

Personalized and precision medicine tend to get conflated these days in the world of branding, but for our purposes, the difference isn't meaningful. Both tailor treatment to the individual patient's characteristics (e.g., lifestyle, genes). It's what practicing doctors do every day; branding lags behind a longstanding clinical reality.

Geisinger's rhetoric describing DNA sequencing misleads what the technology is capable of today, and likely even in the near future. "We are preventing disease from happening" is inaccurate because few diseases have a single cause or purely genomic basis. Currently, this technology is an unfocused screening tool creating anxiety and uncertainty regarding relative risks that may never come to fruition – patient's emotional energy is focused on probabilities.

"We think this will decrease spending on a lot of care" is a guess at best. Screening of the general population, rather than targeted groups results in unnecessary confirmatory testing, over diagnosis, and interventions due to the knee-jerk human reflex to do something instead of nothing. Casting wide nets to satisfy intellectual curiosity rather than clinical concerns can beget unnecessary actions, some that can do harm. Consider prenatal testing that provides probabilities, often false, that lead to irreversible terminations. Of course, there is utility in actionable prevention when risk factors are known, but seeking the right testing is essential to promulgating useful data.

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"For these patients, precision health care reduces uncertainty and allows them to take charge." To date,

valuable precision medicine involves targeted therapies based on a patient's tumor tissue analysis and its genetic variants (for diagnoses that are already known). Is knowing you might be at risk of heart disease or cancer and should, therefore, eat nutritiously and exercise progressing care? Only if you consider these recommendations a new revelation.

Let's address what wasn't said

Geisinger is both a health system and health insurance company. Is there a firewall between these functions for this data, or can it be used by actuaries to alter premiums and coverage? And while the data is stored within your health record who is the owner? Is Geisinger, like the direct to consumer companies planning on licensing or selling genomic information? That puts their offer to cover the \$300-500 cost of the collection and analysis, in a different and less flattering light. There is a value in contributing your genomic information in the aggregate, it is a contribution that benefits all of us, but will the knowledge derived be the exclusive property of the health system? Whether they actually use the data to prevent disease is speculative, that they use it for predictive modeling of their expenses is not.

Finally, there is the toll on the busy primary care physician, already over scheduled, overworked and burdened with data entry. The "What ifs" that result from unfocused genomic information squander the diminishing time of office visits, often to the detriment of the problems that are actually taking place. And while Geisinger is offering its primary care physicians a half-hour video on interpreting this new information, is that sufficient? They also claim the information that can't be acted upon will be withheld, does this not violate the physician's duty to his patient to provide fully informed consent?

As the ever present allure of data for data's sake preoccupies all facets of our culture, only time will tell if these and other questions will soon be answered. Hopefully the drive to acquire good, informative data that does no harm will prevail (see [here](#) on the fallacy of "testing for everything").

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