

The Genomic Revolution: Tying Discovery to Care

Tasha Fingerlin, Ph.D.

Every day, it seems, scientists discover a new genetic health risk. A recent Internet search uncovered reports of genes that raise the risk for cannabis dependency, stroke and being neurotic. The Precision Medicine Initiative seeks to use an individual's genomic profile and other characteristics to develop personalized strategies for preventing and treating diseases with fewer side effects and better outcomes.

This kind of work has already led to discoveries with profound impact on human health. Mutated versions of the BRCA1

gene can raise a woman's chance of developing breast cancer from one in eight to one in two. A particular mutation in the EGFR gene can make an intractable lung cancer susceptible to a treatment targeted specifically at that mutation. Mutations in the APOL1 gene that are found almost exclusively in African-Americans partially explain racial differences in end-stage kidney disease and survival rates of kidney-transplant patients. That kind of information can drive crucial health care decisions.

We must remember, however, that most of the genetic health risks you read about are statistical findings. Biostatisticians, epidemiologists and bioinformaticians generate and search through massive datasets containing billions of pieces of genomic information, looking for subtle patterns associated with health and disease. When a particular piece pops out above the random background noise, we report the association.



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A few years ago, my colleagues at National Jewish Health, led by Dr. Max Seibold and Dr. David Schwartz, reported that a common variant of a gene known as MUC5B is associated with as much as a 20-fold increased risk of developing the fatal and incurable disease idiopathic pulmonary fibrosis (IPF). At present, with no approved treatment that can prolong the life of IPF patients, knowing about that increased risk does not alter an individual's clinical care or disease outcome.

However, the discovery spurred further biological investigations that revealed how MUC5B contributes to excess mucus in IPF and to the study of new medications to treat IPF

by breaking up that mucus deep in the lungs. That approach would never have been considered if not for the original statistical discovery of a genetic risk factor. There are many other diseases for which novel therapeutic strategies are being developed that are motivated by genomic discoveries.

The key is to recognize that the discovery of genetic risk factors is not the end of the story; it is the foundation for understanding the basis of disease. To take true advantage of the genetic discoveries likely to come out of the Precision Medicine Initiative, we need to invest heavily in studies that translate these promising discoveries into more effective prevention and care. Ultimately, these long-term investments in precision medicine will not only improve health, but may reduce the overall cost of care.

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*One in a series of commentaries by leading physicians and researchers at **National Jewish Health** in Denver, Colorado. National Jewish Health is in New York City at the **Mount Sinai – National Jewish Health Respiratory Institute**.*