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FIRST OPINION

Personalized medicine: We're not there yet

By Edward Abrahams and Christopher J. Wells

When Francis Collins, then-director of the National Human Genome Research Institute, testified before Congress in 2003 about the significance of sequencing the human genome that year, he introduced personalized medicine as a new concept. He predicted that in 10 years personalized medicine would allow physicians to employ "predictive genetic tests … so that each of us can learn of our individual risks for future illness and practice more effective health maintenance and disease prevention."

Collins didn't stop there. By the year 2020, he opined, "gene-based designer drugs are likely to be available for conditions like diabetes, Alzheimer's disease, hypertension, and many other disorders. Cancer treatment will precisely target the molecular fingerprints of particular tumors, genetic information will be used routinely to give patients more appropriate drug therapy, and the diagnosis and treatment of mental illness will be transformed."

From a scientific perspective, some of Collins's predictions have come true. In the two decades since his testimony, scientists and industry leaders in personalized medicine have brought forth the promise of big benefits for some people. But many of those products remain out of reach for too many people. For this reason, as leaders of the nonprofit Personalized Medicine Coalition, we are committing an increased share of the organization's resources to eliminating the barriers that people face in accessing the tests and treatments underpinning personalized medicine.

20 years of progress

Because scientists have made so much incremental progress along the lines Collins laid out in 2003, it is easy to miss the validity of his central contention: genomic medicine can improve lives and make health systems more efficient.

Twenty years ago, advocates for personalized medicine had little to show for their enthusiasm. The Food and Drug Administration had approved only a few genetically targeted drugs, though these approvals did provide proofs of principle.

Herceptin (trastuzumab), Iressa (gefitinib), and Gleevec (imatinib) had only recently become available for certain people with breast, lung, and blood cancers. The drugs worked by disrupting the activities of rogue proteins, produced as a result of specific genetic mutations that were known to promote cancer growth.

Early proponents of personalized medicine saw that technological breakthroughs had unlocked unprecedented opportunities to study the activities of genes, proteins, and mRNA, among other molecules. Scientists predicted that an "explosive growth in our knowledge of genetics and the molecular origins of disease" would someday provide the capacity to detect conditions in advance of observable signs of disease. They anticipated new medical interventions that could delay disease onset or minimize symptom severity.

Their enthusiasm was not misplaced: Since 2003, researchers and industry leaders in health and medicine have driven at least 100,000 molecular tests and 350 molecularly targeted medicines to the market. More than half of all clinical trials in cancer care are now for drugs that are targeted to patients with certain genetic characteristics.

Those tests and treatments have delivered — and will continue to deliver — unprecedented opportunities to make good on the promise of personalized medicine: to prescribe medicines for the right patient at the right time and to detect diseases at earlier stages to preempt their progression. Some tests promise to upend cancer care, for example, by diagnosing the disease before any symptoms appear and may be more easily treated.

Today, blood tests can find early-stage cancers. Genomic tests can more quickly pinpoint devastating genetic conditions and their causes. Genetically engineered immune cells are giving cancer patients long-lasting immunity against would-be relapses. And a new wave of gene therapies, which are designed to introduce new genetic material that will counteract disease-causing genetic variants, could potentially cure some patients with rare genetic diseases, including infants and small children whose progressive conditions would otherwise kill them after a few years of extreme suffering.

A widening practice gap

Unfortunately, the rapid pace of scientific progress in personalized medicine has contributed to a widening

gap between what is possible and what is practiced in modern medicine. In a paper published in 2022, researchers showed that genetically based tests and treatments were reaching fewer than 40% of the people with advanced non-small cell lung cancer who may have benefitted from them. For multiple reasons, the other 60% of patients have gained little from the advancement of personalized medicine.

Consequently, as humanity approaches what has been called a "golden age for medicine," the public's questions are rightfully less about the scientific possibilities that will soon be available at academic medical centers and more about the practical realities that shape how most people gain access to the most effective medical interventions.

Who will be able to get blood-based early cancer detection tests? Can the health system afford to spend more on genetic and genomic sequencing tests? And how will economically marginalized people gain equitable access to gene therapies that hold the greatest promise for them?

To make good on America's investment in the Human Genome Project and hundreds of billions of dollars spent on research and development in the private sector, we recognize that the Personalized Medicine Coalition and other advocates for personalized medicine must provide compelling answers to these and other questions related to the clinical adoption and implementation of personalized medicine.

Building a better system for personalized medicine

We and our colleagues at the coalition do not concede, as some critics have argued, that the existence of questions about affordability and access provides a compelling reason to turn away from a scientific endeavor that has brought sight to genetically blind children, cured people with terminal cancer, and helped others avoid late-stage cancer diagnoses that would have meant almost-certain death.

At a systemic level, spending more money on even the most expensive gene therapies, for example, makes sense because the treatments can eliminate the costs of future medical care and hospitalizations that occur when unchecked rare genetic diseases wreak havoc in people. If business leaders and policymakers fail to envision ways for health systems to absorb up-front spending in pursuit of these kinds of downstream savings and cures, they will fail the very people their work is designed to serve.

It is incumbent upon this generation of health care and life sciences leaders to do the hard work that will be required to imagine and build a better health system that prioritizes more institutional spending on the most cost-effective prevention and treatment strategies while protecting people from increased out-of-pocket costs. Scientists and industry leaders can take the lead in this endeavor by devoting a greater share of their resources toward implementation science studies. Such studies focus not on demonstrating the scientific and clinical promise of new products and technologies, which has been the subject of the last two decades of research in personalized medicine, but on understanding how to address the barriers that make it difficult for many people to access them.

Last month, for example, a PMC-led commentary article introduced potential solutions to clinical practice gaps in personalized medicine. This is the beginning of a major workstream in clinical adoption that will also include the development of a roadmap for addressing clinical practice gaps in personalized medicine. In the future, partnerships between our organization and clinical care providers will be designed to demonstrate what can be done to make personalized medicine more accessible to patients.

The science underpinning personalized medicine demands continued investment 20 years after Collins's testimony before Congress. But it is against the yardstick of access that the success of the field will ultimately be measured.

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