Banking on health

Biomedical research and healthcare has traditionally centered on disease rather than health. Several projects gathering data on healthy people promise to change that.

UK Health Secretary Matt Hancock recently announced that the National Health Service, the UK's nationwide health system, will offer healthy individuals in England the option of paying to have their genomes sequenced in return for information on their risk of disease. The move was not universally welcomed. While providing sequence information to patients with hard-to-treat cancers and rare diseases has clear benefits, the same is not immediately true for healthy individuals. Even so, the UK efforts to shift healthcare closer to health reflect a change that can be seen in other large-scale initiatives across the world. The promise—as yet unrealized—is to create a foundational human dataset that will help governments and the research community better to understand what "health" means over a lifetime and how that is related to disease.

Last December, the UK reached its goal of sequencing 100,000 genomes of National Health Service patients. According to Genomics England, the effort has already resulted in one in four participants with rare diseases being given a diagnosis for the first time, and hundreds of cancer patients have been prescribed more tailored treatments.

The same month, the UK government released its Life Sciences Sector Deal 2 report outlining a strategy for "transforming the prevention, diagnosis and treatment of chronic diseases by 2030." Among the key initiatives detailed in the report is a proposal to enable "genomic volunteers" to pay for their genome to be sequenced, returning a "personalised report on their unique genetic makeup" while also making consented data available to the research community.

Gathering genetic information on healthy people is not new. In 2016, for example, the first results were released from the Scripps Translational Science Institute's Wellderly study, which sequenced and analyzed the genomes of over 500 disease-free people over the age of 80. Another example is the partnership between Geisinger Health System and Regeneron to collect samples from more than 100,000 consented Geisinger volunteers. The Regeneron Genetics Center performs sequencing and genotyping to generate de-identified genomic data for drug discovery; following validation of variants in CLIA-certified clinical laboratories, Geisinger then passes the information back to participants.

One challenge—as is evident from marker-driven services like 23andMe—is that genome information only leads to actionable information in a few cases. The medical community is concerned about being inundated with enquiries about variants of unknown significance. Genomic profiling may even result in overtreatment. Thus, despite its critical importance in advancing human biology research, the likelihood of large numbers of healthy people participating in sequencing remains limited, at least for now.

That said, an increasing range of efforts are aiming to capture a much more comprehensive set of human health data.

Since 2006, the UK Biobank has enrolled 500,000 people aged 40 to 69 for a combination of genome sequencing and measurements of behavior, social life, mental state, lifestyle, diet and physical health. Overall, data on more than 2,400 different traits or phenotypes have been collected. Full genotyping data were released on the entire cohort of 500,000 in 2017, and later this month 50,000 exome profiles will be publicly released. According to the UK Biobank, 7,000 researchers have registered to access the data.

In the United States, the 100K Wellness Project has completed its first phase of massive profiling that included over 1,000 markers in a cohort of 108 healthy people tracked for 9 months, which has now expanded to > 5,000 individuals. The All of Us Research Program is also gearing up, with plans to gather molecular, lifestyle and environmental measurements on a million or more people (http://allofus.nih.gov/), and the Kavli Foundation's HUMAN Project is setting out to track the behavior of 10,000 New Yorkers for the next 20 years.

In Asia, the Wellcome Trust–funded China Kadoorie Biobank has gathered data on 515,000 people from the Chinese mainland. And last year, ~$90 million was earmarked to fund the first phase of the International Human Phenome Project, which has plans to build a phenotypic map and database of around 1,000 healthy Chinese individuals (ultimately growing to 10,000).

These efforts are notable because they contrast with the 'disease' focus of biomedical research, an approach grounded both in the structure of medical specialties and in the need to address 'indications' that trigger action from physicians and payments from reimbursers. Research institutions, public (the US National Institutes of 'Health') and private (biopharmaceutical R&D), are transfixed by illness. Vaccination aside, healthcare and medicine is organized around late-stage symptomatic disease.

This disease myopia restricts both the type of funding and cross-disciplinary expertise needed to enable human health profiling.

The good news is that the economics of population health may be changing. Healthcare systems are beginning to consolidate health provision and payment. As evidence one need only look at a recent study from Providence St. Joseph Health detailing the benefits of prioritizing interventions designed to correct underlying pathophysiological disturbances (i.e., impaired insulin secretion and resistance) in individuals at high risk for diabetes. Similarly, visionary employers are beginning to engage in health research and population health (for example, the recent partnership of Amazon, Berkshire Hathaway and JPMorgan Chase).

Understanding health matters. We can only comprehend inflammatory disorders or autoimmunity if we know what is meant by a 'healthy' immune system. We can only understand mental health if we understand the nervous system before the onset of symptomatic disease. We can only design interventions to alter gut microflora if we know what a 'healthy' microbiome looks like.

Companies like Arivale, Human Longevity, Verily, Amgen/deCode Genetics, Regeneron and iCarbonX understand this. And as analytical technology, smart phone technology, wearables, soft electronics and ultimately implants become more miniaturized and unintrusive, the ability to carry out massive phenotyping in a regular or even continuous manner on healthy individuals promises to transform human research. It heralds an era where we can finally begin to collect the data needed to better understand what health, in all its manifestations, really is. ❐

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