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# The Human Digitome: a 21st-century 'omics to deliver personalized medicine

Digital trial company Medable has created the first Human Digitome, a digital platform that represents human health and disease and is built to provide real-world data for personalized and predictive medicine.

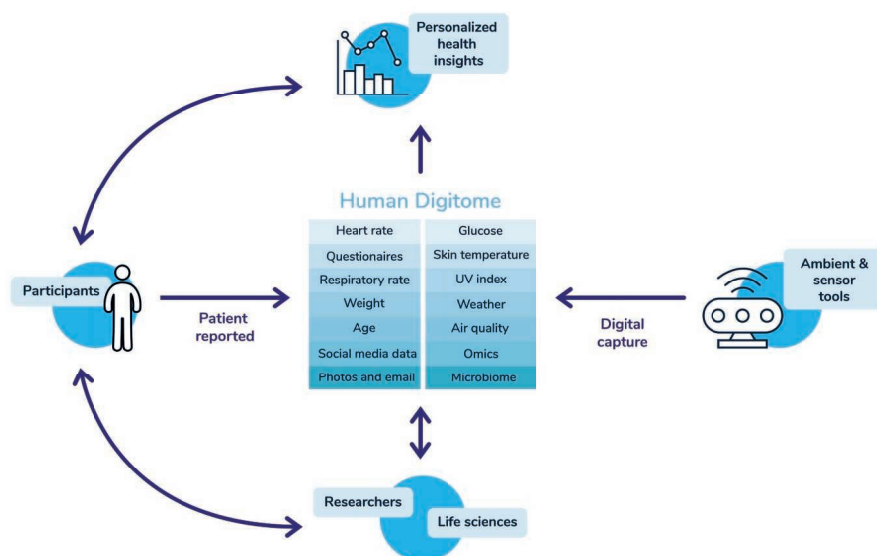
Medable, the Palo Alto-based digital health provider, is developing the world's first comprehensive digital representation of human health and disease, to drive personalized and predictive medicine. Since its launch in late 2016, Medable has seen rapid adoption of its platform with a customer base that cares for more than 15 million patients. While digital tools can enable scientific discovery, it costs \$2 billion and 12 years in time to bring a new drug to market and only 1 in 5,000 drugs developed effectively ever makes it to the market. Using digital device data collection, Medable has enabled researchers to study disease on a greater scale with a larger population sample and more representative patient health data.

By aggregating all these data, Medable built the first Human Digitome, a digital representation of human health and disease, which provides foundational real-world data for digital biomarkers, therapeutics and population-level research insights.

The scientific community envisioned great medical advances from the Human Genome Project, the natural culmination of the history of genetics research. This international, collaborative research program, for which the goal was the complete mapping and understanding of all human genes, contributed to significant scientific progress, and the full sequence was completed and published in April 2003.

The impact of sequencing and mapping of the human genome has yet to be fully realized in terms of lives saved and diseases prevented. In an era in which an individual's genome can be sequenced for as little as \$1,000—the price point that, it was surmised, would usher in a new era of personalized health and medicine—why is it still elusive? Clearly, genomics plays an important role in disease, but there are other external factors that have an impact on clinical outcomes and individual health, and persistent data silos inhibit rapid discovery.

To truly 'decode' the human genome for medical insights—that is, to understand how slight differences in genetic sequences amongst the world's population give rise to differences in disease risk, disease manifestation and treatment outcomes—the medical community needs complementary high-resolution phenotypic data of individuals and populations, across health and disease, collected continuously over time and available in a unified dataset. Until very recently, capturing this type of data at population level was not feasible. Historically, health data have been divided into silos and captured in sporadic clinical visits that often rely on subjective levels of documentation by individual practitioners, in difficult-to-access 'doctor's notes'. However, in the



**Fig. 1 | The Human Digitome will prove essential to unlock the full potential of preventive interventions and personalized medicine.**

current era of constant data generation, digital data that contain highly relevant information about lifestyle and environmental exposures can be captured continuously outside the clinic. Some of these data that have traditionally not been considered 'medical' may provide missing phenotypic insight into factors that affect physical and mental health. For example, the passive collection and mining of vast amounts of text-based data generated by people every day may help to identify early signs of cognitive decline. By analyzing changes in word choice and sentence structure of an individual over time outside the clinic, practitioners may be able to diagnose certain dementias earlier, or better understand mental health disorders and risk.

The rapid digitization of health care and ubiquitous technologies such as smartphones and wearables that passively collect real-time phenome data provide an opportunity to build valuable datasets. These datasets may be able to help clarify the complex relationships between genetics, environment, lifestyle and disease risk.

## The Human Digitome

These unique and novel data sources are the dawn of 21st-century 'omics—the Human Digitome ("Digitome"), a fully digital representation of human health and disease. Digital data generated by individuals over time represent subtle differences in

phenotypes that have been elusive. Recording and decoding these data offer the potential to rapidly advance the understanding of how the human body functions, why diseases occur, as well as providing insight into prediction and personalized treatments. As participant-level data-sharing becomes the new norm in clinical research and patients demand better access to their own health records and information, there is an opportunity to accelerate scientific discovery, compile more robust data and investigate previously unanswerable questions. Academic researchers, clinicians, patients and pharmaceutical companies conducting clinical trials all aim to better understand health and provide health-care solutions (Fig. 1).

"Defining and characterizing the Human Digitome is going to be one of the most important endeavors for the life sciences in the 21st century," said Jacob Laporte, cofounder & VP, global head of The Novartis Biome. "Predicting human health, disease risk, progression and therapy response, based on an individual's genetic information and environment is like a very complicated cryptography problem. In the 20th century, we made significant strides in characterizing the code through genomics, proteomics and transcriptomics. However, scientists have struggled to develop a uniform key to decode this information into nuanced insights for a given individual's health. One reason behind this is the lack of rich, longitudinal phenotypic information. As the digital health



# HUMAN DIGITOME

revolution kicks into high gear, we have the opportunity to capture this missing piece of the puzzle and really make unprecedented progress in realizing true personalized medicine.”

## The Human Digitome is driven in part by the Internet of Things

With the rise of the Internet of Things, a connected world of devices and objects that collect and exchange data is being seamlessly embedded into everyday life. Within homes and bodies, multifaceted digital data streams are being generated that are unique to each individual. When this information is combined with digital social interaction data and electronic health records, a rich picture emerges that holds insights into an individual's mental and physical health.

The Internet of Things extends beyond consumer devices to a rapidly growing market of medical devices that remotely capture human physiology, outside the clinic or hospital. The volume of data captured is truly unprecedented. Health measurements of the future will no longer be limited to singular categories such as heart rate or temperature, but instead will represent time-bounded composite measurements that take into consideration novel physiological parameters, traditional vital signs and external factors such as air quality and medications.

As sensors become increasingly sophisticated, they will collect multifaceted measurements of an individual's overall health (including physiological and behavioral factors) in a single device (for example, Alexa) that ambivalently senses with no individual burden. For the first time, it may be possible to elucidate multidimensional relationships such as subtle sleep parameters that combine with air quality to affect cardiac function or induce asthma attacks.

The potential to record and improve health based upon personalized Digitomes is remarkable. Health

professionals may rapidly pinpoint individuals with risk factors for specific illnesses as well as understand disease progression to create more impactful and timely interventions. For example, in today's world, health professionals often lack enough information about an individual to accurately characterize stroke risk and intervene in a preventive manner. Most people see their primary care physician once a year or less, and the data collected during these visits are limited and cannot be used to predict stroke risk. Using individual Digitomes, it may be possible to discern whether an individual is likely to suffer a stroke or even a heart attack, before it happens. For example, in the Cardiogram, Inc. study conducted in conjunction with the Division of Cardiology, Department of Medicine at the University of California San Francisco, researchers used Apple Watch data collected from a population of patients to passively detect atrial fibrillation (AF), using a deep neural network. The research team used this digital data to create a neural network algorithm. They then showed that they could use this algorithm to passively detect AF in a different population, paving the way for early detection and AF intervention using specific digital data.

## The Human Digitome ultimately stands to revolutionize medicine

The world is in the midst of a digital revolution, where public demand for people's own health-care data converges with current advancements in machine learning and artificial intelligence (AI) to develop new screening techniques and medical treatments to radically improve human health and well-being. However, to synthesize the data and navigate the complexities of various disease 'omics, it is necessary to begin capturing and mapping the Human Digitome to create and leverage agreed-upon methods and standards. The Human Digitome will

enable preventive screening techniques, similar to the one developed in the Cardiogram study, as well as more accurate disease models to maintain and improve human health. “Within the Digitome, we are using AI to look for patterns that detect the transition between health to disease. We believe the Digitome is only at about 1% of what it could be—and in the next few years we are planning to open up Medable's platform so that patients can proactively use it themselves, adding their data to the Digitome to help researchers understand how diseases work, and how to fight them,” said Michelle Longmire, CEO and cofounder of Medable.

The multidimensional nature of the Human Digitome will enable advances in earlier disease prediction, detection, and ultimately cures. Advanced analytics will unlock the significance of common and rare genetic variants to fulfill the promise of the Human Genome in connection with lifestyle and environmental exposures. Now is the time to move forward and build the Human Digitome with multiple scientific disciplines. The Human Genome Project is an important initial step and milestone in the understanding of human health and disease, but to deliver and unlock the full potential of personalized medicine, the Human Digitome is an essential component for the future of patient care.

For more information, how to engage, and the progress of the Human Digitome, please contact [digitome@medable.com](mailto:digitome@medable.com).

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