Human 'molecular map' contributes to the understanding of disease mechanisms

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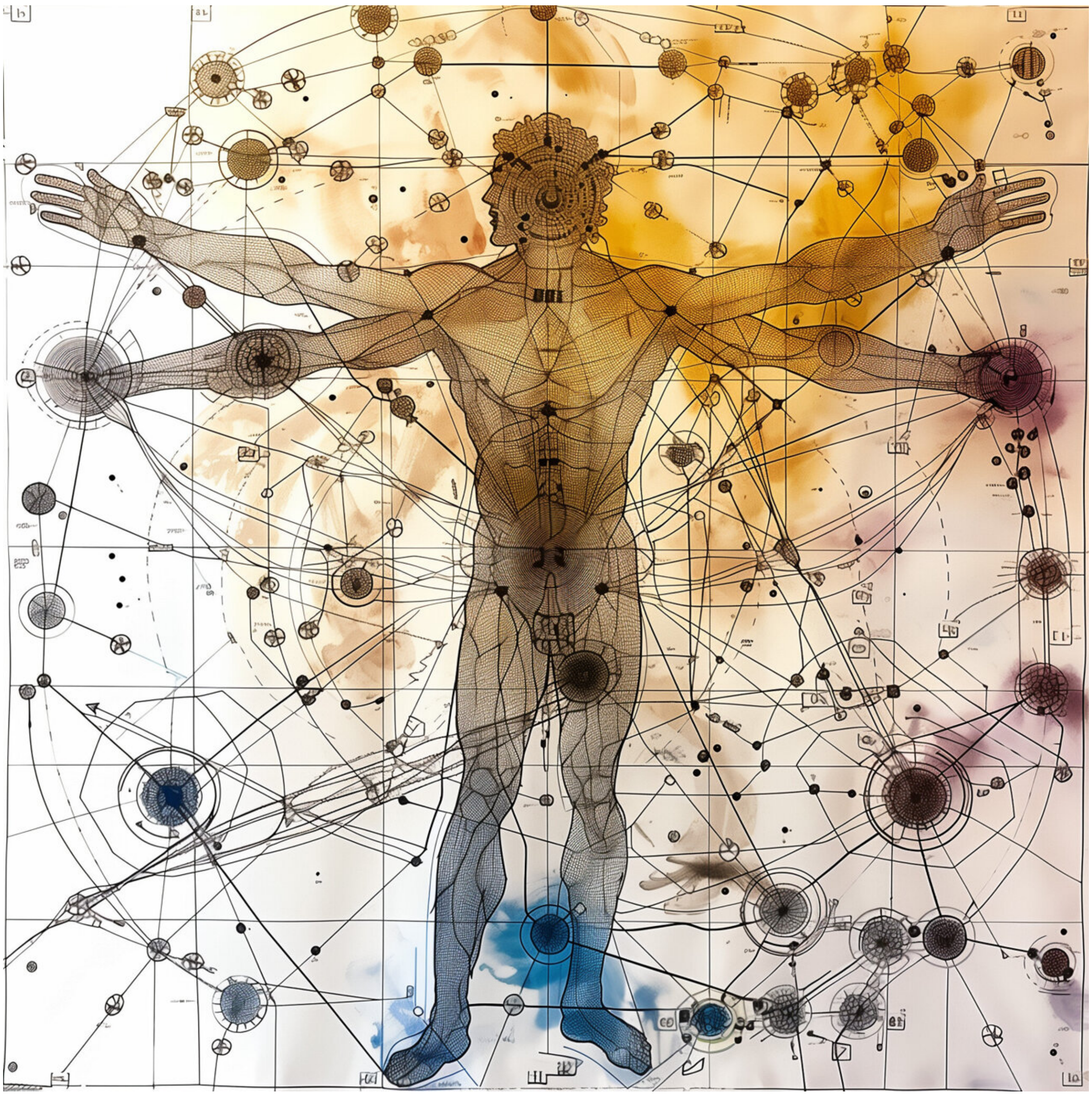

Researchers at Weill Cornell Medicine in Qatar have created a free-to-access online reference tool that scientists around the globe can use for investigating how the human body works at the molecular level and forming hypotheses to test with experimentation. Credit: Suhre/Halama Labs

Scientists at Weill Cornell Medicine in Qatar (WCM-Q) have created an intricate molecular map of the human body and its complex physiological processes based on the analysis of thousands of molecules in blood, urine and saliva samples from 391 volunteers. The data was integrated to create a powerful, interactive visual web-based tool called Connecting Omics (COmics) that can be used to investigate the complex molecular make-up of humans and discover underlying traits associated with various diseases.

The molecular processes of the human body refer to the chemical reactions and interactions occurring within cells and between different cells, including crucial functions like DNA replication, protein synthesis, energy production, cellular communication and various metabolic pathways, all governed by complex protein-protein, protein-DNA, and protein-RNA interactions, ultimately enabling the body's vital functions.

The exhaustive study, published Aug. 19 in *Nature Communications*, collated 12 years of data from the Qatar Metabolomics Study of Diabetes (QMDiab), a diabetes case-control study in the multiethnic population of Qatar, predominantly Arab, Filipino and Indian backgrounds.

"Our idea was to bring together everything we have learned over more than a decade of multiomics research to create a comprehensive molecular model of the human body and its processes," said senior author Dr. Karsten Suhre, professor of physiology and biophysics and a member of the Englander Institute of Precision Medicine. "This reference tool is free to access and use by researchers who want to investigate how the human body works at the molecular level and also for the formation of hypotheses to test with experimentation."

Through a collaboration with Hamad Medical Corporation, the researchers collected multiple aliquots of blood, urine and saliva samples from volunteers, with and without diabetes. The samples were subsequently characterized on 18 different high-throughput analysis platforms, providing an extremely rich dataset including 6,300 individual molecular data points including genomic data (DNA), transcriptome (RNA), proteins and metabolites, such as amino acids, sugars and fats. In addition, they determined information on genetic variants, DNA methylation sites and gene expression for each of the participants.

This allowed the researchers to discover associations and pathways linking genetic characteristics with specific proteins, metabolic processes and diseases. They then painstakingly integrated the mass of data from all the individuals into an online web-based tool serving as the interface to "The Molecular Human," the molecular description of the human body.

The approach of combining genomic, transcriptomic, metabolomic, proteomic and other forms of so-called -omics research is known as multiomics. This approach has emerged in recent years as a key strategy for biomedical researchers seeking to understand how the human body and diseases truly function, providing insights that could potentially enable the development of new drug therapies.

For instance, the study identified and described the proteins and metabolites which are signatures of subtypes of type 2 diabetes, shedding light on the different ways the disease manifests.

"Our integrative omics approach provides an overview of the interrelationships between different molecular traits and their association with a person's phenotype—their observable traits, such as their physical appearance, biochemical processes and behaviors," said first author Dr. Anna Halama, assistant professor of research in physiology and biophysics.

"The scale of the data integrated within the COmics web tool enables access to hundreds of thousands of pathways and associations for researchers to explore, giving huge potential for discovery and investigation."

More information: Anna Halama et al, A roadmap to the molecular human linking multiomics with population traits and diabetes subtypes, *Nature Communications* (2024). DOI: 10.1038/s41467-024-51134-x

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