Study of metabolism-related genetic factors reveals new associations between blood biomarkers and diseases

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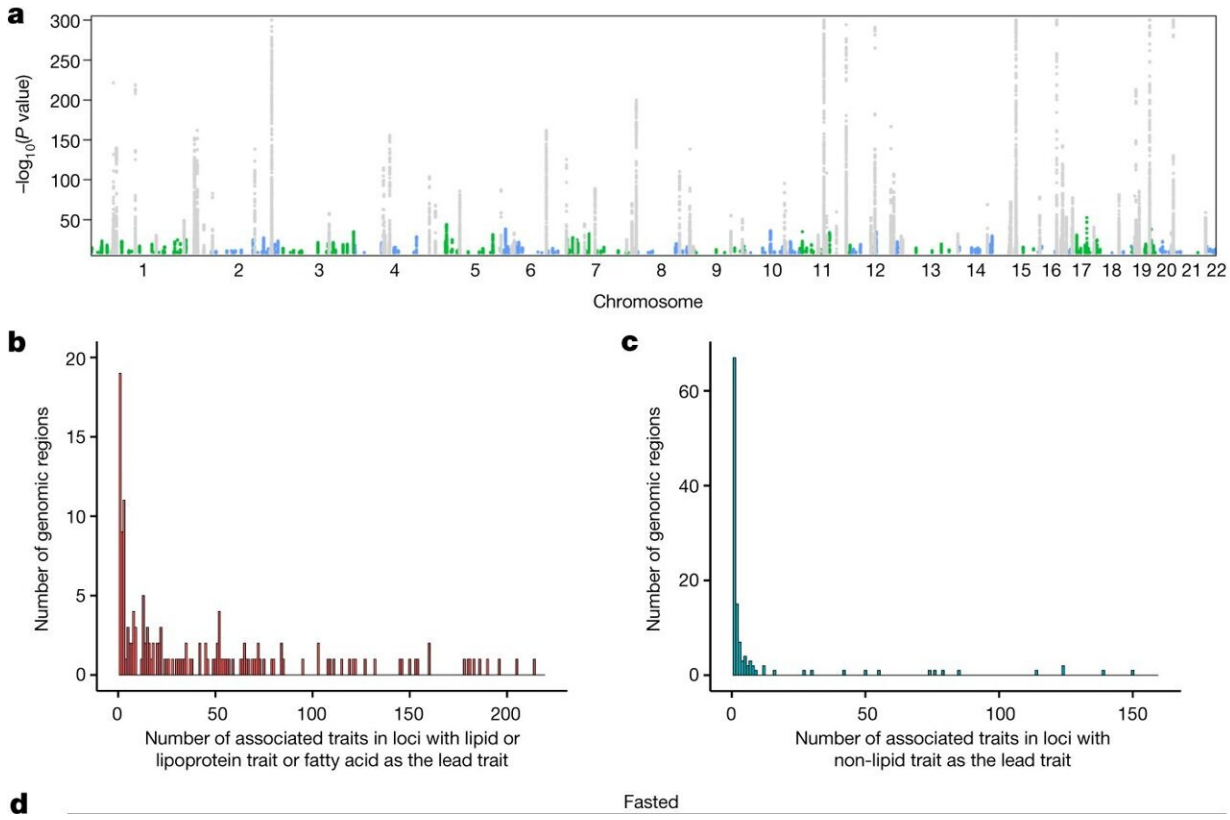

Results of the GWAS meta-analysis of 233 metabolic traits. a, Manhattan plot summarizing the metabolic trait associations from inverse variance-weighted GWAS meta-analysis. b,c, Numbers of associated metabolic traits at the 276 associated genomic regions are shown separately for genomic regions in which the lead trait was a lipid, lipoprotein or fatty acid trait. d, Results of the GWAS for glucose for the fasted cohorts. Credit: *Nature* (2024). DOI:

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An international study led by the University of Oulu, Finland, and the University of Cambridge, U.K., found numerous associations between blood biomarkers and diseases, and identified more than 400 genomic regions that affect metabolic regulation. Many of these findings are completely new. The results were published in *Nature* on March 6, 2024.

The study found that a large proportion of the genomic regions related to metabolism are also linked to the risk of developing various diseases. The new research results help to detail the biological processes through which blood markers are linked to diseases. The results can be used, for example, in the development of new medications and in clarifying the cause-and-effect relationships between metabolic biomarkers and diseases.

The combination of 33 population datasets enabled the study in 136,000 volunteers. Six Finnish population cohorts were in a key role in the study, including the Northern Finland Birth Cohorts, housed at the University of Oulu. Additional data were used from the UK Biobank, which allowed confirmative analyses in 120,000 participants. The researchers analyzed 233 metabolic biomarkers from blood samples and examined their connections to more than 13 million genetic factors.

"We learned a tremendous amount of new biology about the heritable factors of human metabolism and their associations with diseases," says one of the principal researchers, adjunct professor Minna Karjalainen from the University of Oulu.

For example, the study described the metabolic effects of those genetic factors that predispose to intrahepatic cholestasis of pregnancy. These

new findings shed light on this rather common, but not well-understood, liver disorder diagnosed in about one in a hundred pregnant women.

"The study is an excellent example of the key role of genetics in understanding metabolic pathways in relation to various diseases," says Professor Johannes Kettunen from the University of Oulu. He is an expert in metabolism-related genetics and is one of the leaders of the new study.

The study applied state-of-the-art metabolic analyses, based on nuclear magnetic resonance (NMR) spectroscopy, that provide more than 200 metabolism-related biomarkers from a single blood sample. This methodology is also very cost-effective and therefore suited well for the studies of large population datasets. Professor Mika Ala-Korpela, from the University of Oulu, has led the development of this methodology in collaboration with the NMR Metabolomics Laboratory at the University of Eastern Finland, Kuopio.

"This study is a splendid demonstration of the central importance of new research methods—in this case, both metabolic and genetic analyses—and their multidisciplinary combination to develop science and to reveal the intricacies of nature," Ala-Korpela says. He is one of the leaders of the study. He founded the Computational Medicine research team, focused on metabolism and genetics, at the University of Oulu, Faculty of Medicine, in 2009.

The key results of the study are openly available to the scientific community and will enable a plethora of follow-up studies. The main authors of the study work at the University of Oulu and at the University of Cambridge, U.K.

More information: Minna K. Karjalainen et al, Genome-wide characterization of circulating metabolic biomarkers, *Nature* (2024).

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