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Sidra Medicine establishes Middle Eastâ??s first atlas of DNA variants shaping genome regulation

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Nature Genetics study creates a new resource to unravel genome complexity and advance precision medicine for Arab populations



Researchers at Sidra Medicine, a member of Qatar Foundation, have created the Middle East's first population-scale atlas showing how genetic variation regulates gene activity in blood and tracing its effects to clinical traits.

Published in Nature Genetics, the study, titled "A catalogue of transcriptome genetic regulation in a consanguineous Middle Eastern biobank", provides a new resource to advance precision medicine and addresses a major gap in global genomics and advances precision medicine by enabling more precise genetic diagnosis and disease-risk prediction, while supporting the discovery of new biomarkers and therapeutic targets.

Despite major genome sequencing efforts worldwide, little is known about what most genetic variants do and how they influence health and disease. This gap is even greater for Arab and Middle Eastern populations, where genetic diversity and the broad effects of high consanguinity remain understudied.

The research represents a significant milestone in improving the interpretation of genomic test results and increasing the yield of genetic diagnosis through better annotation of non-coding variants. Also, it enables the development of ancestry-aware, functionally-informed polygenic scores for better assessment of risk to complex diseases and more successful clinical trials. By linking known variants to their target genes, the findings will also prioritise new targets for functional investigation and the validation of biomarkers and therapeutic targets grounded in disease biology.

The study was supported by Sidra Medicine and the Qatar National Research Fund, leveraging data and samples from Qatar Biobank and the Qatar Genome Program, now part of the Qatar Precision Health Institute.