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Toward a fine-scale population health monitoring system

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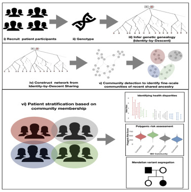
Highlights

- Genomic data linked to health records capture demography in health systems
- Genetic networks reveal recent common ancestry in diverse populations
- Evidence of many founder populations in New York City
- Fine-scale population structure impacts genetic risk predictions

Summary

Understanding population health disparities is an essential component of equitable precision health efforts. Epidemiology research often relies on definitions of race and ethnicity, but these population labels may not adequately capture disease burdens and environmental factors impacting specific sub-populations. Here, we propose a framework for repurposing data from electronic health records (EHRs) in concert with genomic data to explore the demographic ties that can impact disease burdens. Using data from a diverse biobank in New York City, we identified 17 communities sharing recent genetic ancestry. We observed 1,177 health outcomes that were statistically associated with a specific group and demonstrated significant differences in the segregation of genetic variants contributing to Mendelian diseases. We also demonstrated that fine-scale population structure can impact the prediction of complex disease risk within groups. This work reinforces the utility of linking genomic data to EHRs and provides a framework toward fine-scale monitoring of population health.

Graphical abstract


i) Recruit patient participants
ii) Genotype
iii) Infer genetic genealogy (Identity-by-Descent)
iv) Construct network from Identity-by-Descent Sharing
v) Community detection to identify fine-scale communities of recent shared ancestry
vi) Patient stratification based on community membership
Identifying health disparities
Polygenic risk assessment
Mendelian variant segregation

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Keywords

electronic health records • computational genomics • genomic medicine • machine learning • biobanks • genetic ancestry • population health • health disparities

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