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Using population biobanks to understand complex traits, rare diseases, and their shared genetic architecture

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Propositions

1. Large-scale population-based biobanks are powerful tools to make accurate inferences about the data that is not included in their collections (this thesis).
2. COVID-19 disease severity is independent from the common genetic variation of the human cellular receptors ACE2 and TMPRSS2 used for SARS-CoV-2 entry (this thesis).
3. Integration of different levels of phenotypic and genetic information from general population provides novel insights into the mechanism behind incomplete penetrance of rare mutations, with potential for clinical applications (this thesis).
4. Host genetics partially affect the microbiome composition and function, and these determinant genetic loci are important for host metabolism and health (this thesis).
5. The increase of genetic diversity in population biobanks greatly improves the yield of genetic discoveries (this thesis).
6. Association of individual common variants from well powered studies is replicable across populations if proper harmonization of phenotyping and methodology is achieved; however this is not true for polygenic scores (this thesis).
7. Researchers building polygenic scores in multi-ancestry studies should evaluate and fine-tune the parameters, accounting for the architecture of the trait and the ancestry, LD patterns and allele frequencies of the target population (this thesis).
8. It is necessary to start building studies, frameworks and data references for non-European populations, but it is urgent to do it for African populations.
9. Even after harmonization, data-set heterogeneity is still a major confounder for population and ancestry-specific effects.
10. Negative results are just as important as positive ones and need to be valued as such.
11. “Destiny can be framed in a model called deterministic chaos, which has 2 components: one random (stochastic) and one deterministic derived from it” - Gabriel Bedoya Berrío.
12. Studies of population genetics and epidemiology should be visualized as impactful factors in the transformation of society to a knowledge-based equitable civilization.