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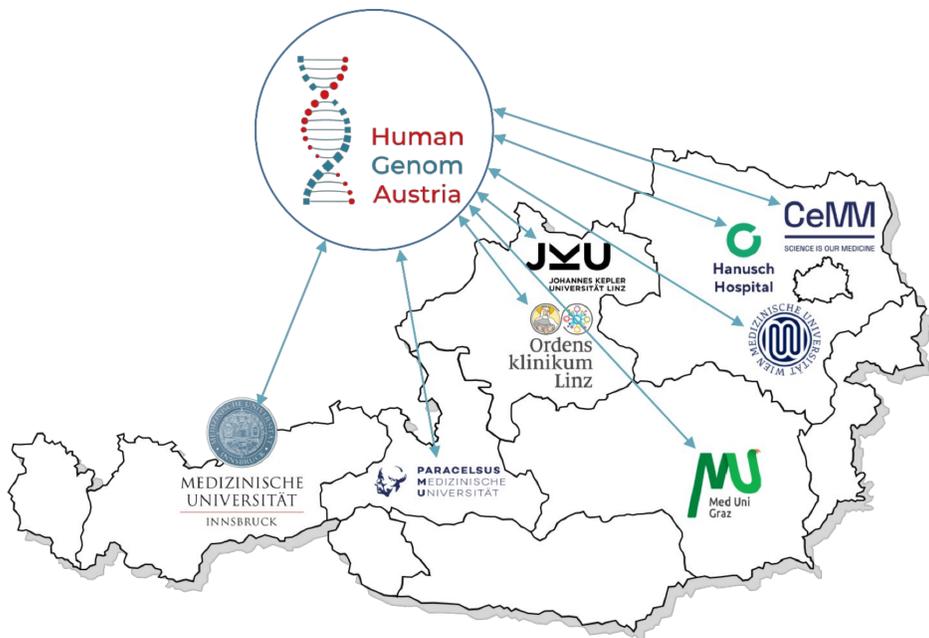
### Background

Austria has been one of the last European countries without a national genome project. Based on the participation in the Genome of Europe initiative, jointly by the Medical Universities of Innsbruck, Graz, and Vienna, and the Center for Molecular Medicine in Vienna, we are now establishing a national genome infrastructure for Austria.

### Take Home

The Austrian Human Genome Project will

- establish a critical genetic reference for the Austrian population
- strengthen collaboration between genomic centers in Austria and Europe
- enhance healthcare and research capabilities in Austria.

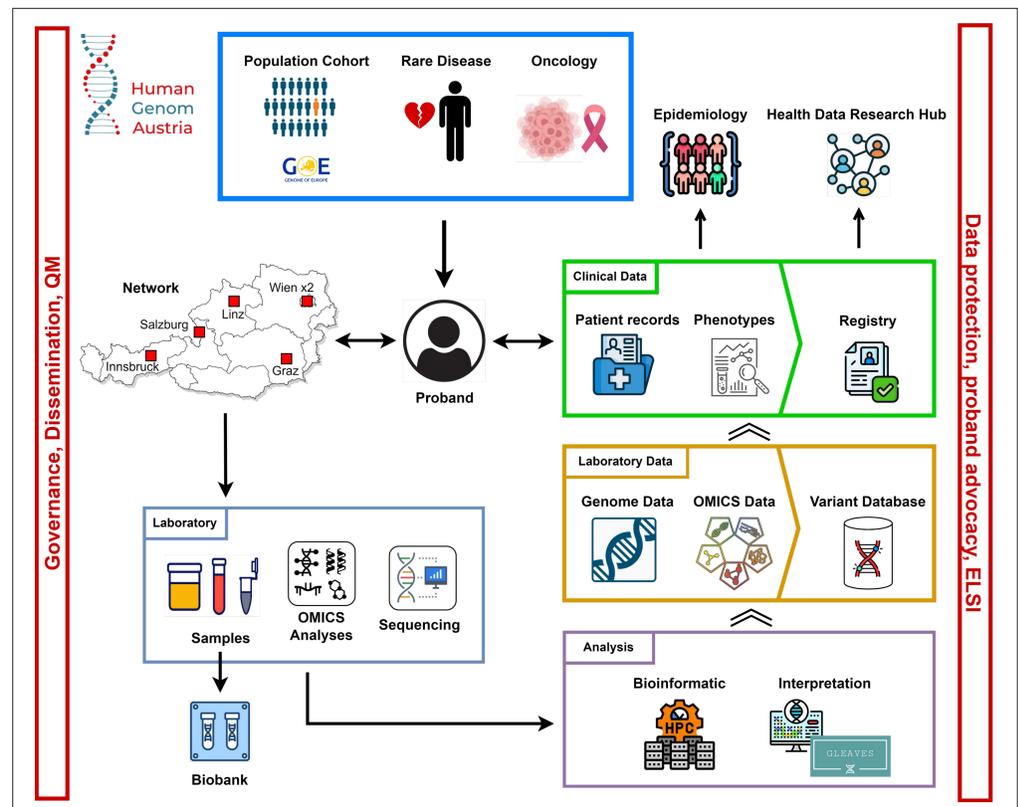


### Objectives

The uses cases addressed by the Austria Human Genome Project will be

- the establishment of Population reference cohort with more than 2150 representative healthy participants (contribution to Genome of Europe)
- the integration of rare disease diagnostics at the six Centers for Medical Genetics in Austria
- the creation of an infrastructure for tumor sequencing and analysis.

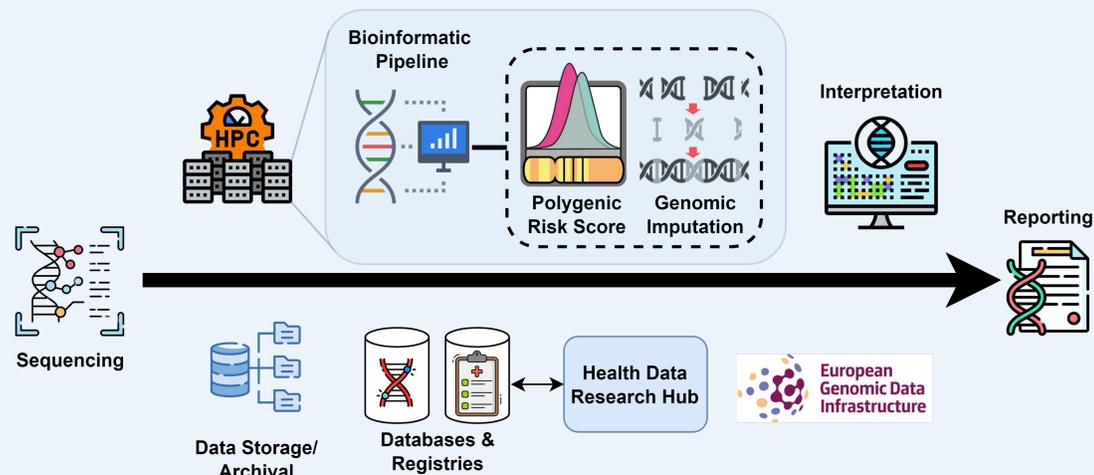
Sequencing will be performed at all participating centers. A state-of-the-art centralized computational infrastructure including data management, analysis, and interpretation pipelines will be established in collaboration with international partners.



### Planned Outcomes

- The primary outcome of the project will be disease, variant, and phenotype databases, registry facilities, a proband and patient access portal, and as well as computational services supporting interpretation, genotype imputation, and polygenic risks scores.
- The infrastructure will connect Austria's medical genomic centers, including two ISO 15189 accredited sites, and will be integrated into European genomic and health data networks.
- The prototyping of the bioinformatics pipelines utilizing a centralized HPC in Innsbruck has been completed.

### Infrastructure Services



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No conflict of interest to declare.