New study shows potential of UK Biobank to inform genetic disease research

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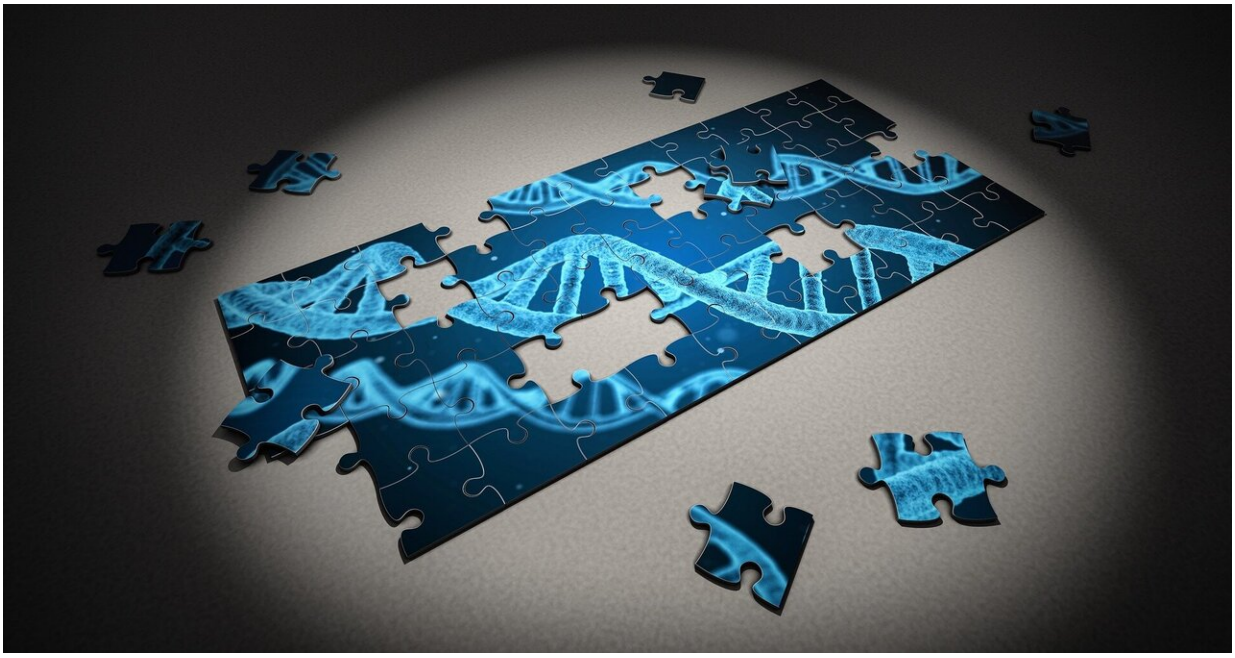

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A new study by RCSI University of Medicine and Health Sciences examining population genetics across Europe has analyzed the diverse ancestries of people living in the UK. This knowledge has the potential to inform future health research on genetic factors leading to disease.

The study, led by researchers at the RCSI School of Pharmacy and

Biomolecular Sciences and the SFI FutureNeuro Research Centre, has been published in *Proceedings of the National Academy of Sciences*.

The RCSI and FutureNeuro researchers used the UK Biobank, a database of genetic and health information of over 500,000 participants from the UK, to examine population genetics and ancestry across Europe.

The study analyzed the genetic ancestry data of individuals in the UK Biobank who reported having a European birthplace outside of the UK—about 1% of the dataset. Researchers catalogued where individuals shared segments of their genome with other individuals, meaning they had a common ancestor within the past 3,000 years.

With this information, the researchers could group individuals with more segments in common than on average into three branches, corresponding to southern, central-eastern, and northwestern Europe.

By studying the patterns of the genome sharing, the researchers were able to infer historical patterns such as population size and how genetically isolated specific European regions are, relative to each other. In general, people from southern Europe were found to have less in common genetically with each other than in other areas, due to the larger population sizes and therefore usually greater number of ancestors in the region.

An exception to this was Malta, which—being an island—was found to have a smaller pool of ancestors. This is the first large sample analysis of Maltese population genetics. Identifying European regions such as Malta with specific histories of genetic isolation could potentially aid the discovery of genetic factors contributing to disease.

In addition to building and expanding upon previous knowledge in

Europe, the results present the UK Biobank as a source of diverse ancestries beyond the UK. This has the potential to complement and inform researchers interested in specific communities or regions across Europe and the world.

Professor Gianpiero Cavalleri, Professor of Human Genetics at RCSI, Deputy Director of FutureNeuro and senior author on the paper, commented, "This research has shown the diversity of European ancestries sampled by the UK Biobank and has enabled us show the 'big picture' of the genetic landscape of Europe, including new insights into communities such as within Malta. This work suggests similar gains of knowledge could be found within non-European ancestry groups using the UK Biobank, groups that are typically excluded from genetic analyses."

Dr. Edmund Gilbert, NUI Posdoctoral Fellow and first author on the paper, commented, "The power of the large sample size of and scale of information on the participants in the UK Biobank has allowed us to show the diversity of genetic histories across the European continent. With new sequencing data becoming available from the UK Biobank, our work lays a foundation for informed analysis of rare and functional variation in ancestries in the UK and beyond."

More information: Edmund Gilbert et al, Revealing the recent demographic history of Europe via haplotype sharing in the UK Biobank, *Proceedings of the National Academy of Sciences* (2022). DOI: 10.1073/pnas.2119281119.

Provided by RCSI

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