Study illustrates unique genetic landscape in Newfoundland and Labrador with links to Ireland and England

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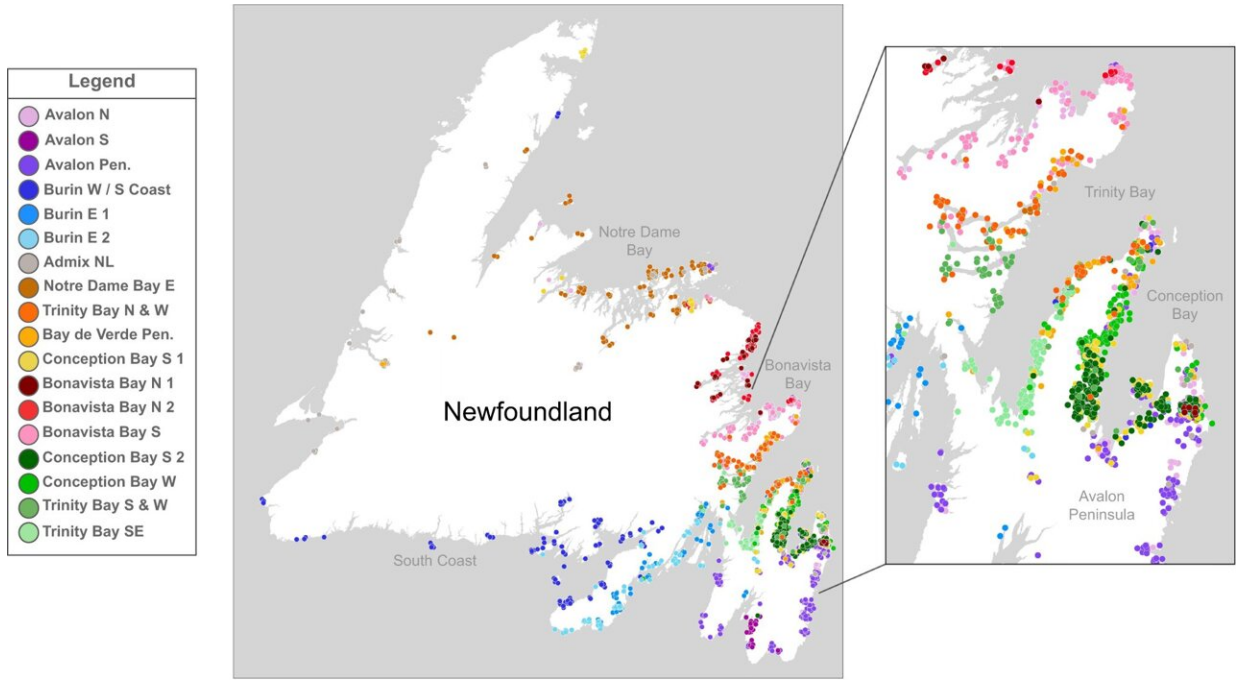

Genetic landscape of Newfoundland and Labrador. Map of the grandparental birthplaces of individuals with color and shape coded according to *fineSTRUCTURE* cluster. A small jitter has been introduced to aid legibility and preserve anonymity. An insert shows individual details of the Trinity and Conception Bays. Panel was plotted within photoshop, with geography boundary data sourced from Tableau. Credit: *Communications Biology* (2023). DOI: 10.1038/s42003-023-04844-9

A new study by RCSI University of Medicine and Health Sciences, based in Dublin, Ireland, and Sequence Bio, a genomics and precision medicine company based in St. John's, Newfoundland and Labrador (NL), Canada, has produced the most detailed genetic analysis of people living in the Canadian province to date, demonstrating a unique founder population structure that could be used for the identification and study of health-related genetic variants.

The study, titled "Newfoundland and Labrador: A mosaic founder population of an Irish and British diaspora from 300 years ago," has been published in *Communications Biology*.

By studying the genetic profiles of 1,807 volunteering individuals from Sequence Bio's Newfoundland and Labrador Genome Project (NLGP), and comparing the resulting fine-scale genetic structure of NL to reference datasets for Ireland and England, scientists showed that a significant proportion of the European-derived population of NL can be traced back to settlers who primarily migrated from South-East Ireland and South-West England around three centuries ago.

"In looking at the ways Newfoundlanders and Labradorians are genetically related to each other, and to present day Irish and English individuals, we were able to show that European ancestry in NL is mainly descended from Irish and English settlers in the time of the late 1700s to early 1800s," explains Dr. Edmund Gilbert, a Lecturer at the School of Pharmacy and Biomolecular Sciences in RCSI and FutureNeuro, the Science Foundation Ireland (SFI) Research Center for Chronic and Rare Neurological Diseases.

Dr. Gilbert, the first author on the study, used well-characterized population reference datasets like the Irish DNA Atlas to link English and Irish ancestry in NL to specific regions in Ireland, and to track how social and geographical isolation influenced NL communities at the level

of their DNA.

Dr. Gerald Mugford, director of research at Sequence Bio commented on the study, "Through this expert collaboration with RCSI, we now have a much deeper understanding of the ancestry of the current NL population and the origins of genetic variants that could be meaningful for disease gene discovery in the province."

Further analysis of the genetic data also shows multiple population bottlenecks, or reductions in population size, happening independently in the region around 300 years ago due to geographical isolation and tendency for people to settle with others from the same country of origin and religious affiliation.

Professor Gianpiero Cavalleri, Professor of Human Genetics at RCSI School of Pharmacy and Biomolecular Science and Deputy Director of the SFI FutureNeuro Research Center, helped lead the comparative study of genomes from Canada, Ireland and England. He said, "The genetic analysis supports the historical accounts that around 25,000 European settlers came to NL in the 18th and 19th centuries, mainly from Ireland—predominantly Waterford, Wexford, south Kilkenny, southeast Tipperary, and southeast Cork—and from Dorset and Devon in England as well as fishing ports such as Dartmouth, Plymouth, or Southampton.

"In the study, we could see that Catholic background in Newfoundland and Labrador is still today strongly associated with Irish genetic ancestry as is Protestant background with English genetic ancestry."

Dr. Michael Phillips, the study's Senior Author commented, "Our findings support NL's population structure as a unique genetic landscape with founder effects." He also noted the potential clinical and health-related importance of these patterns. "Because NL resembles that of

other isolated island populations, there may be an opportunity to study the genetic makeup of specific subpopulations in NL to identify rare genetic variants that contribute to the risk and severity of certain diseases."

More information: Edmund Gilbert et al, The Newfoundland and Labrador mosaic founder population descends from an Irish and British diaspora from 300 years ago, *Communications Biology* (2023). DOI: 10.1038/s42003-023-04844-9

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