

# Reconstructing the genomic history of Eastern Polynesians: insights into phenotypic variation in the Pacific

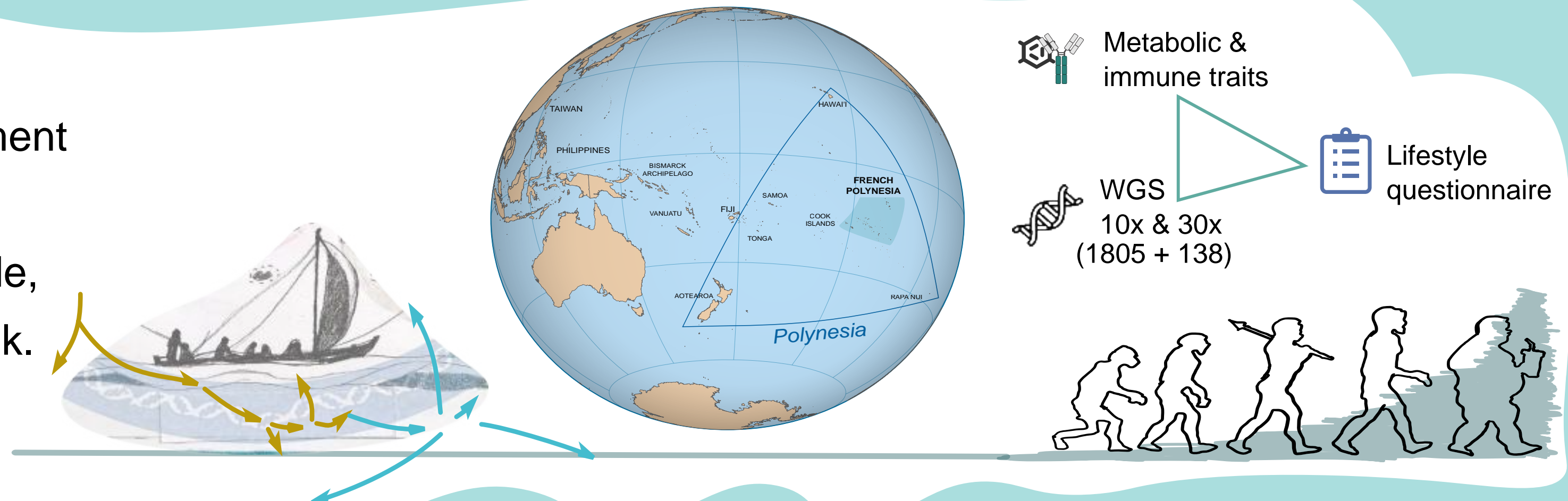
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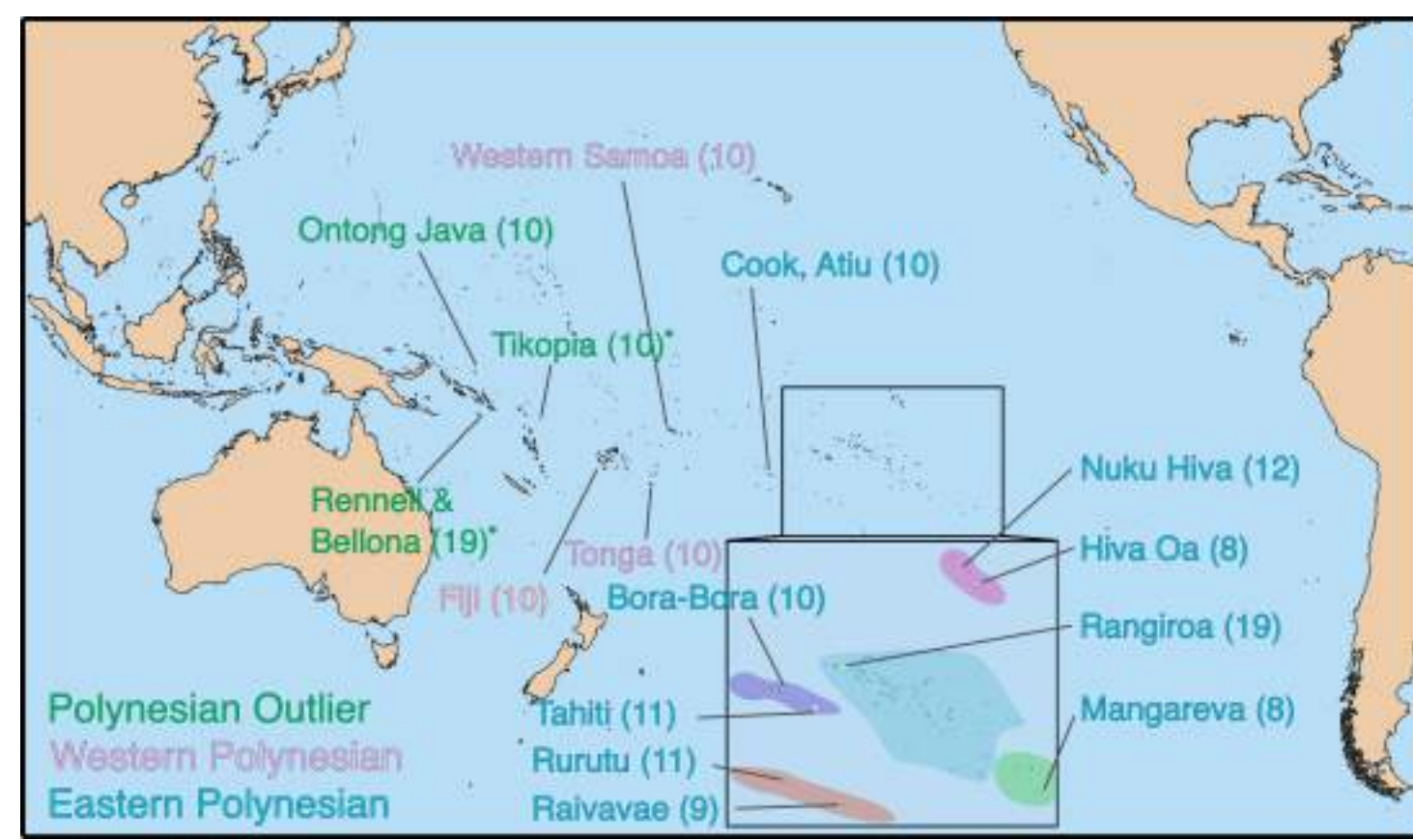
## 1 Introduction

- As the most eastward destination of the Austronesian expansion from Taiwan, the settlement of Polynesia is thought to have involved **extensive admixture and founder events**.
- Polynesians present some of the **highest prevalences of metabolic disorders** worldwide, raising questions about the role of natural selection and genetic drift in their heightened risk.
- The **demographic history** of Polynesians and the **genetic architecture** underlying their phenotypic variation remain largely unexplored.

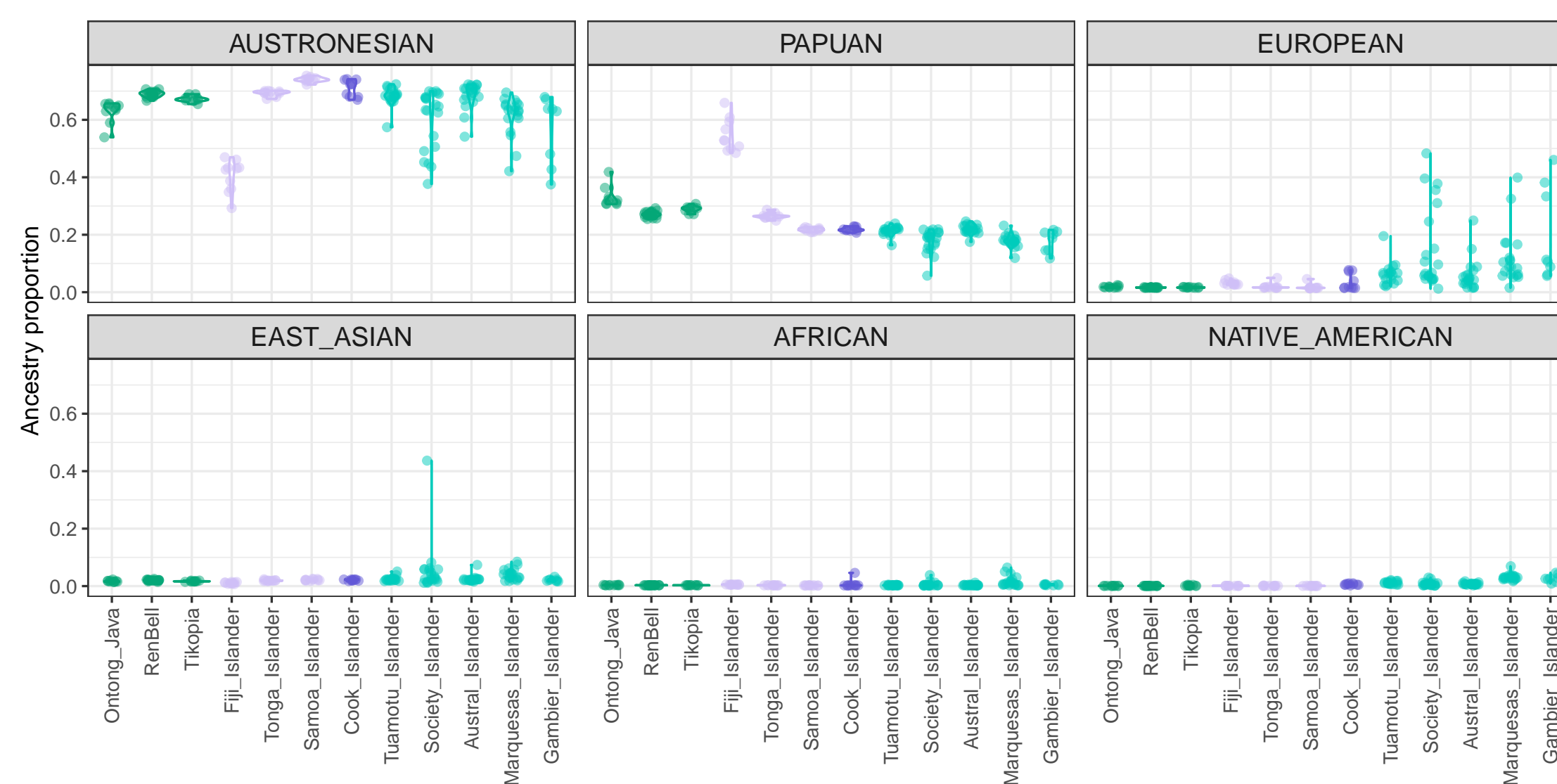


## 2 Demographic history

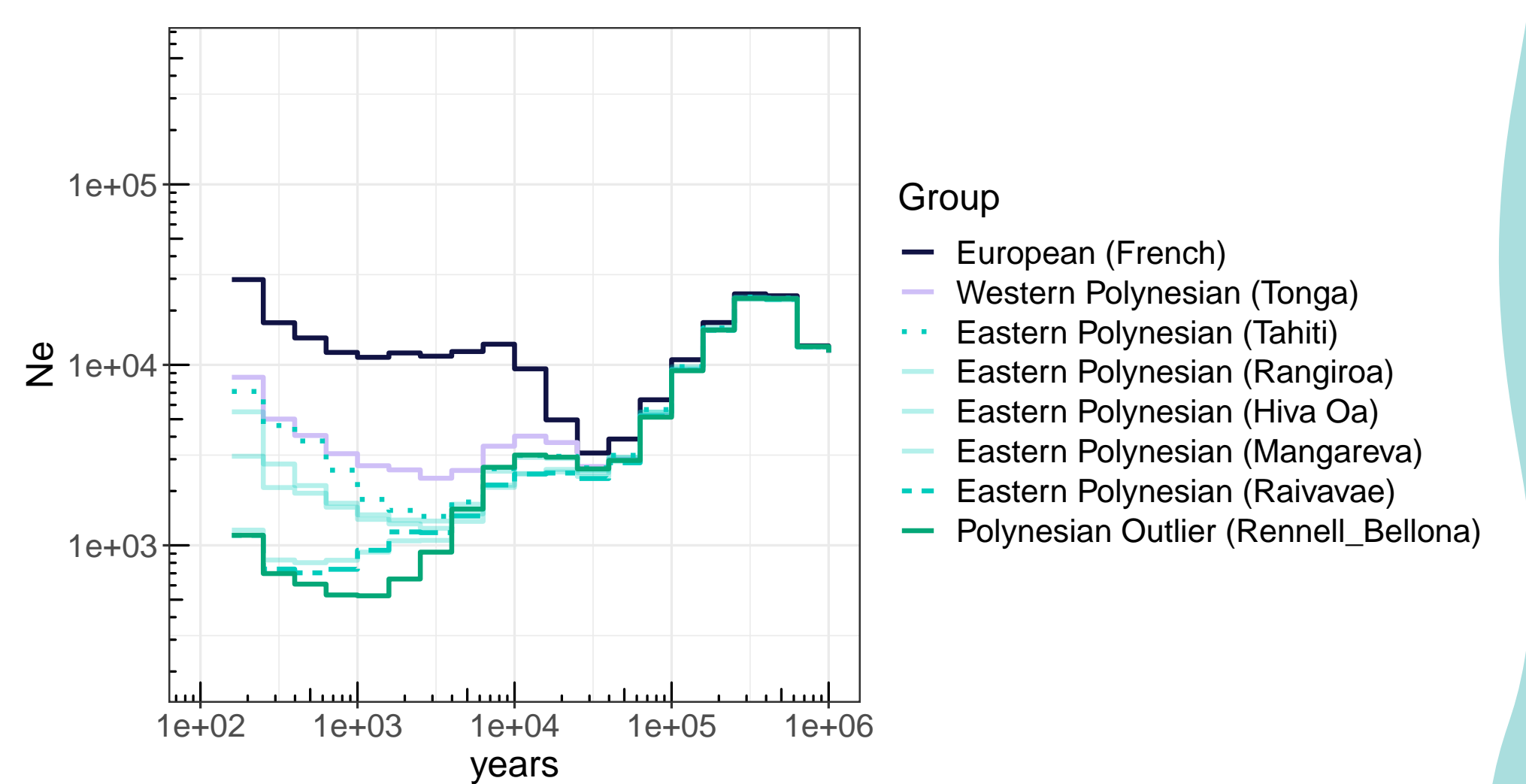
a. Sample map of 138 new 30x genomes



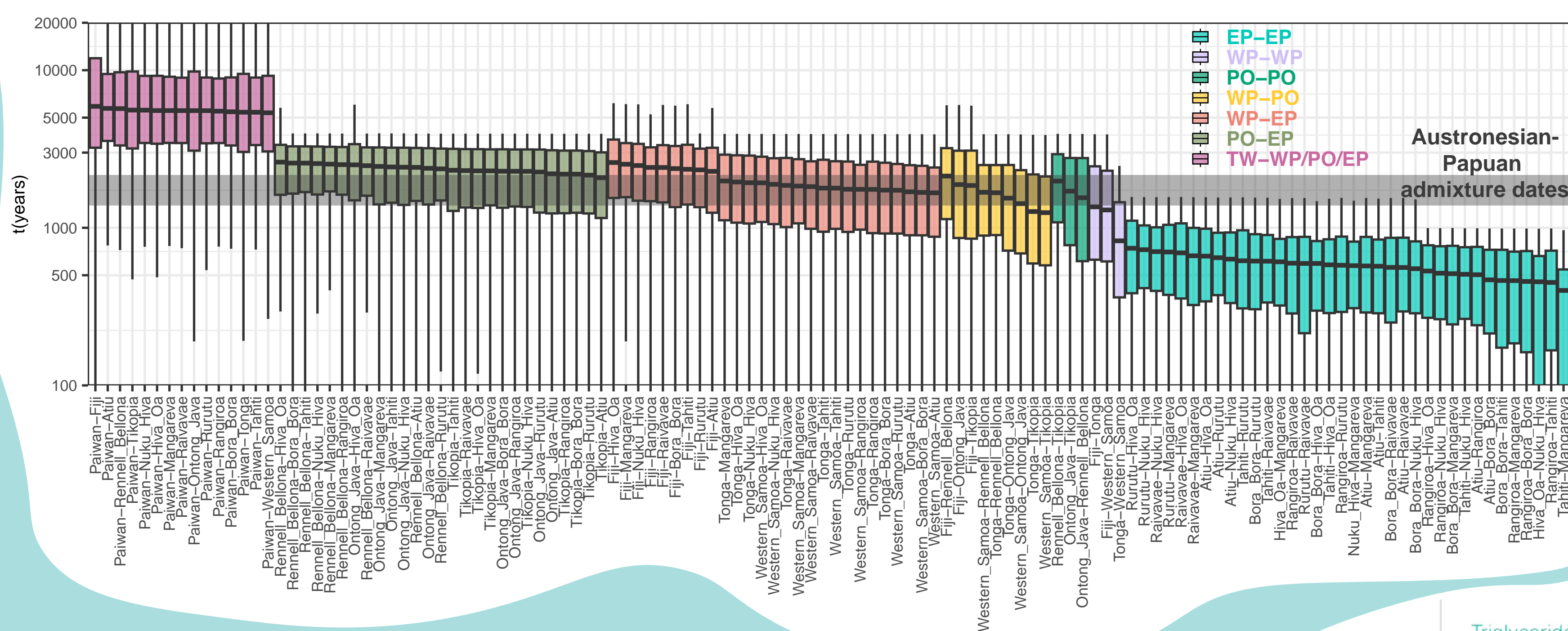
b. Ancestry proportions from summed local ancestry



c. Austronesian ancestry-specific population size of Polynesians, compared to population size of Europeans



d. Austronesian ancestry-specific population divergence

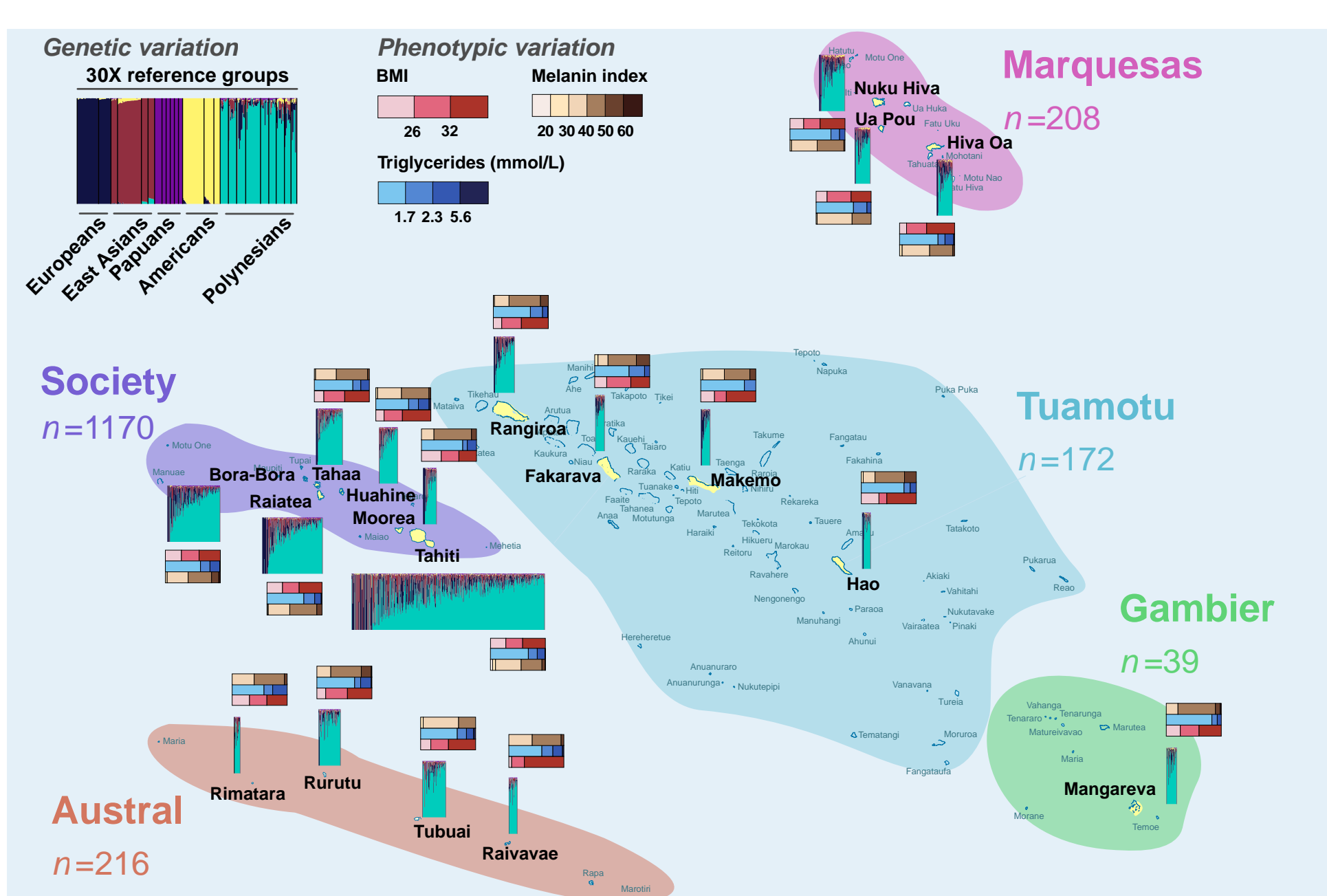


## Conclusion 1:

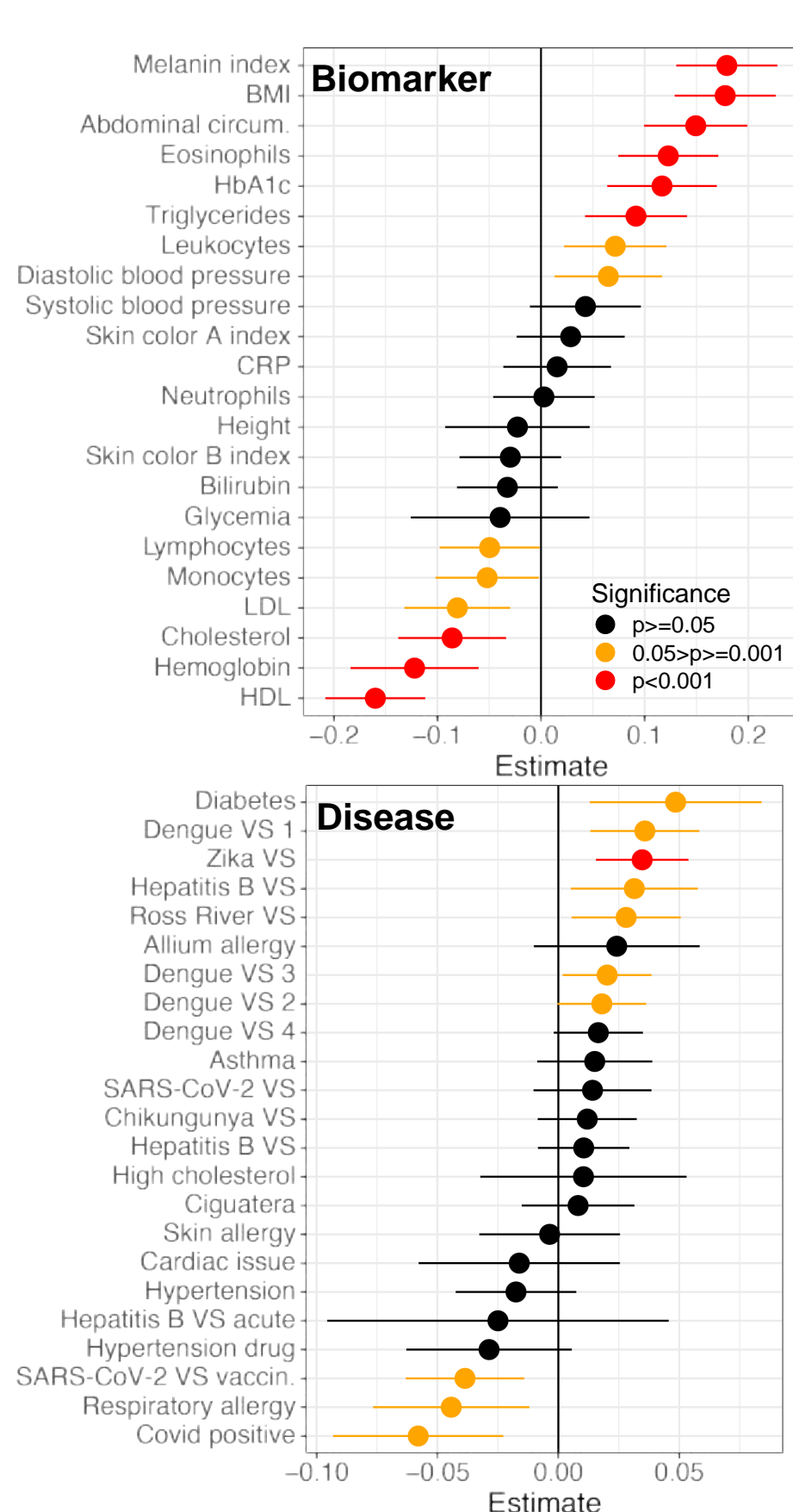
- Polynesians descend from a **major admixture event** between **Austronesian- and Papuan-related groups** dated to **~2.1 kya**, followed by varying degrees of admixture with Papuans, Europeans, East Asians, Africans, and Native Americans.
- A **simultaneous radiation of Polynesians** occurred at **~ the same time**, followed by **strong bottlenecks in Eastern and Outlier groups**.
- Another radiation occurred within **Western and Eastern Polynesia** over the last **~1,000 years**.

## 3 Genetic architecture

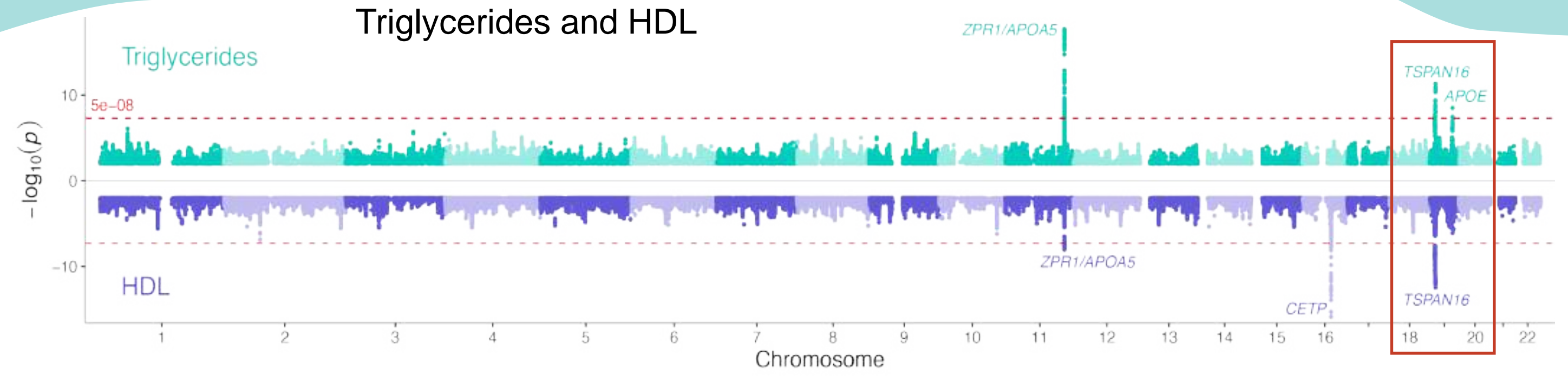
a. Map of genetic & phenotypic variation of 1805 new 10x genomes



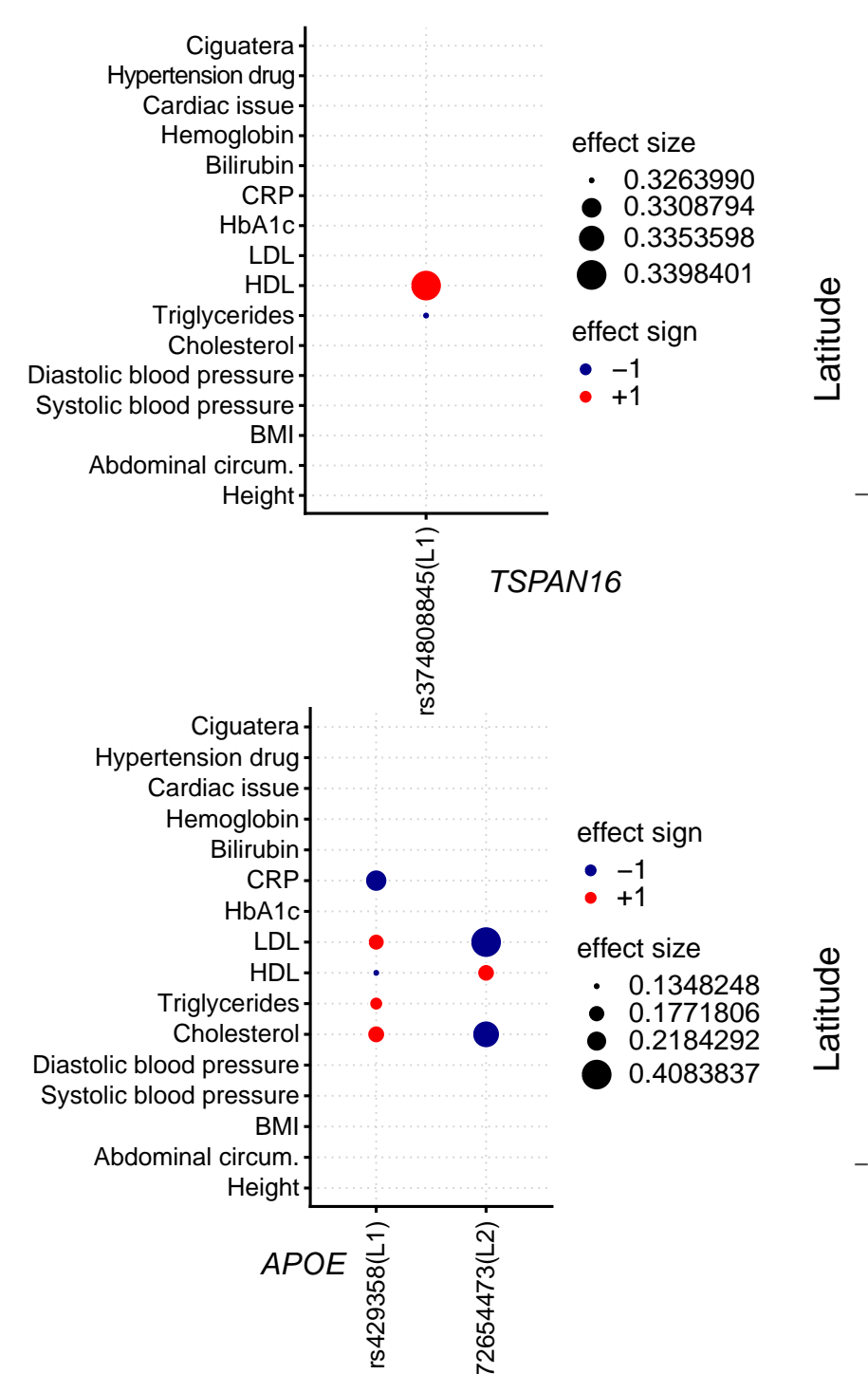
b. Association between genetic ancestry and traits



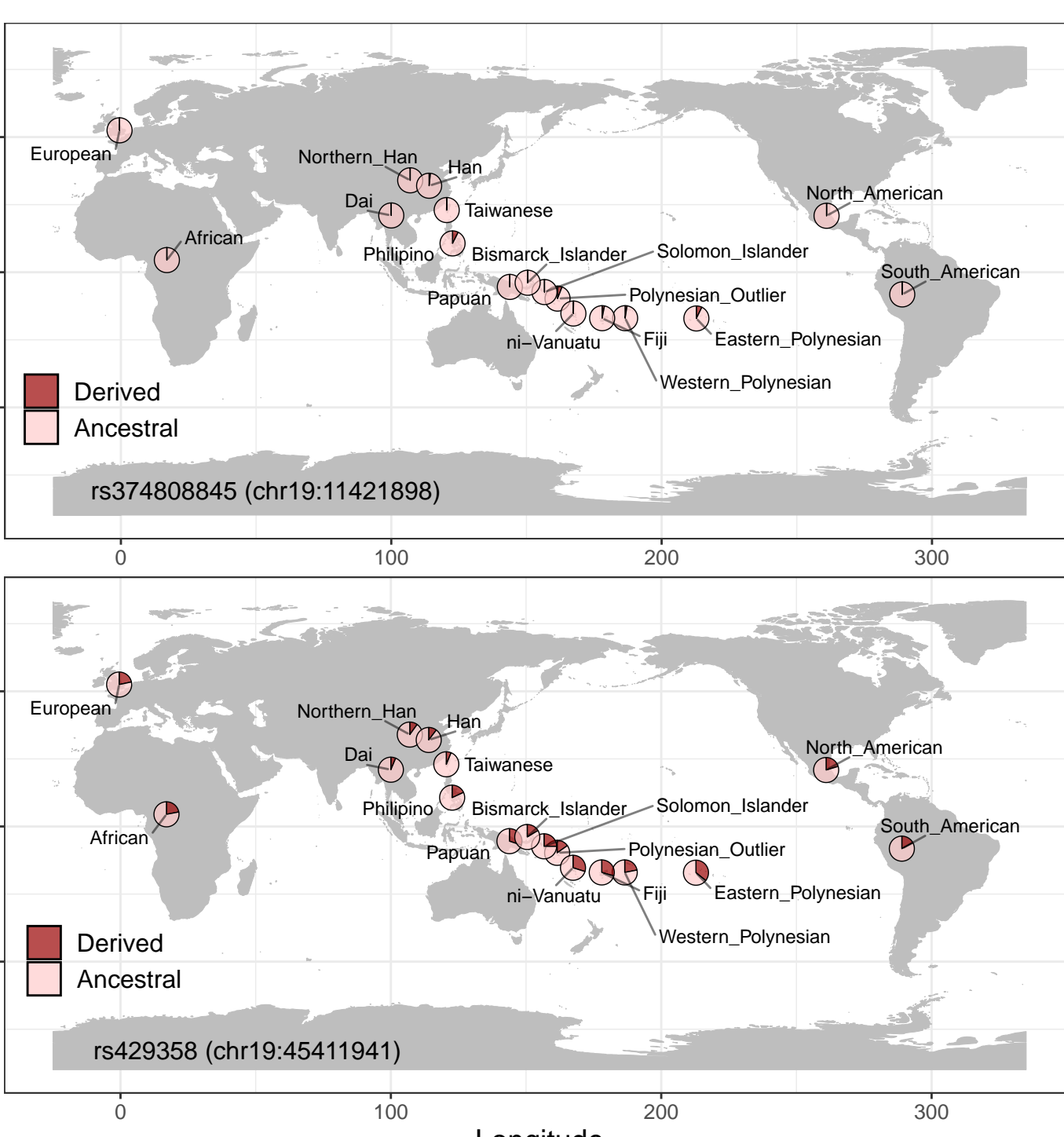
c. GWAS Chicago plot for Triglycerides and HDL



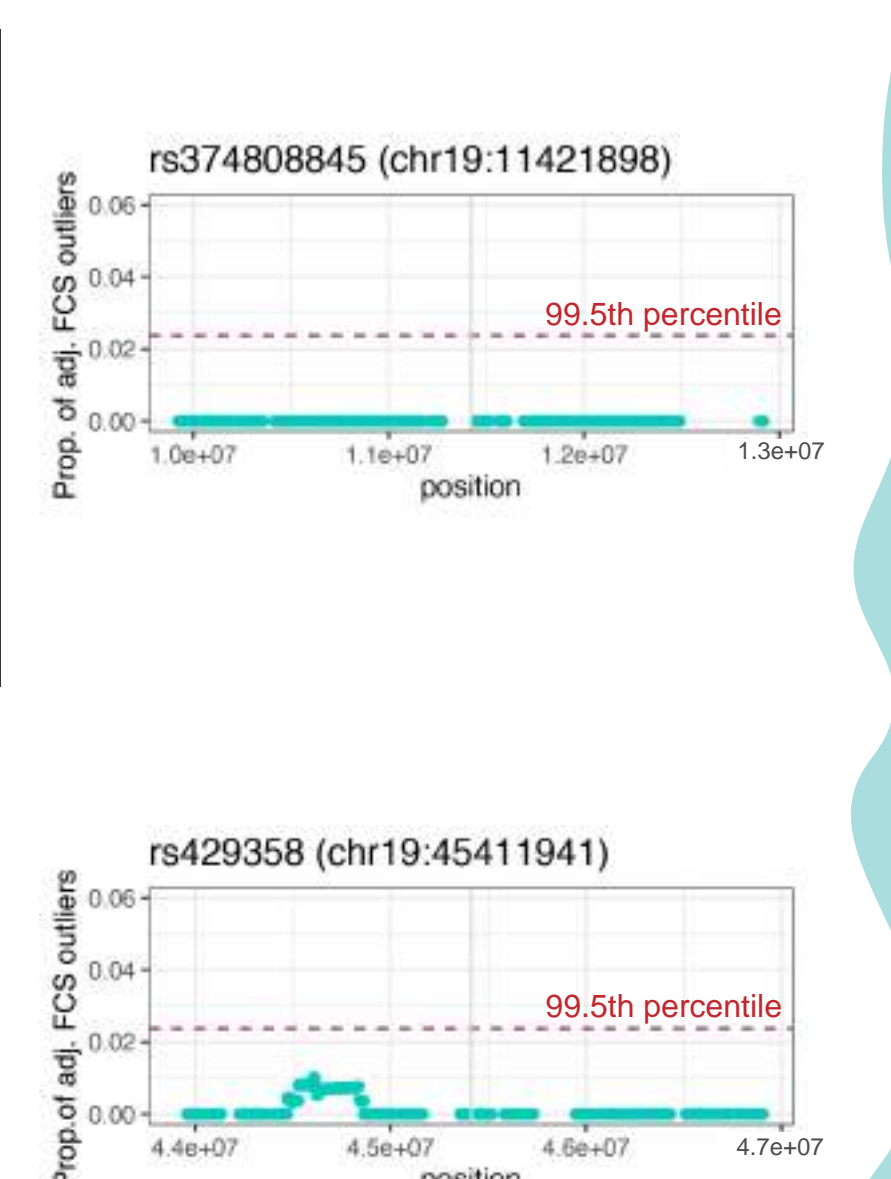
d. Multitrait fine-mapping of the hits at TSPAN16 and APOE



e. Worldwide allele frequency of the fine-mapped hits



e. Selective sweep scan of the fine-mapped regions



## Conclusion 2:

- The **genetic ancestry** of Polynesians is associated with **skin color**, and several metabolic (e.g., **BMI, Diabetes, HDL**) and immunological (e.g., **Zika, Covid**) traits.
- Several variants/genes are associated with lipid traits, including novel hits, such as a variant at the **TSPAN16** locus; the derived allele strongly increases **HDL** and decreases **Triglycerides**, and is **specific (~5-15%)** to in **East Asians and Polynesians**.
- We found **no evidence of strong selective sweeps** at the associated loci, suggesting a **predominant role of genetic drift** in shaping metabolic trait variation in Polynesians.

Scan Me



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