

Characterization of the common genetic variation among the Spanish population of Navarre

Supplementary Information

Supplementary Figures

Figure S1: Accumulative number of new variants contributed by individuals **a)** All common variants in NAVARREsel (21,174 SNVs). **b)** The 676 variants are exclusively common in NAVARREsel and validated in NAVARREval.

Figure S2: **a)** Principal component analysis of overlapped variants between NAVARREsel, MGP, and 1KGP (including all populations), and coloured by superpopulations. **b)** Principal component analysis of overlapped variants between NAVARREsel, MGP, and 1KGP (including exclusively European populations).

Figure S3: Genetic admixture analysis of 1128 individuals from 7 European populations, at an individual level, for the optimal K value = 3.

Figure S4: Comparison of MAF between NAVARREsel and NAVARREval encompassing both validated and non-validated variants, with a total of 998 SNVs.

Figure S5: Minor allele frequency spectrum for the following distinct sets: 21,174 common variants in NAVARREsel (blue colour); 1069 common variants in NAVARREsel but not in *1KGP_noIBS*, gnomAD and *spain* (pink); and 676 common variants in both NAVARREsel (green) and NAVARREval (yellow). Common variants are defined with a MAF > 1%.

Supplementary Tables

Table S1: F_{st} values between Navarre against MGP and 1KGP populations.

Table S2: Variants' information of the 676 exclusively common SNVs of the Navarre population.

Details: "CHR": chromosome; "Position": variant position; "REF": reference allele; "ALT": alternate allele; "NAVARREsel_maf": MAF in NAVARREsel cohort; "NAVARREval_maf": MAF in NAVARREval cohort; "1KGP_phase3noIBS": MAF in 1KGP excluding IBS cohort; "Spain": MAF in *spain* cohort (combining IBS and MGP); "gnomAD": MAF in gnomAD; "Gene": gene where the variant is located; "Type": type of variant; "ExonicInfo": exonic variant type, if applicable; "ClinVar": ClinVar information; "SpliceAI": Splice AI results; "REVEL": REVEL results; "CADD": CADD results; "Polyphen2": Polyphen2 results.

Figure S1

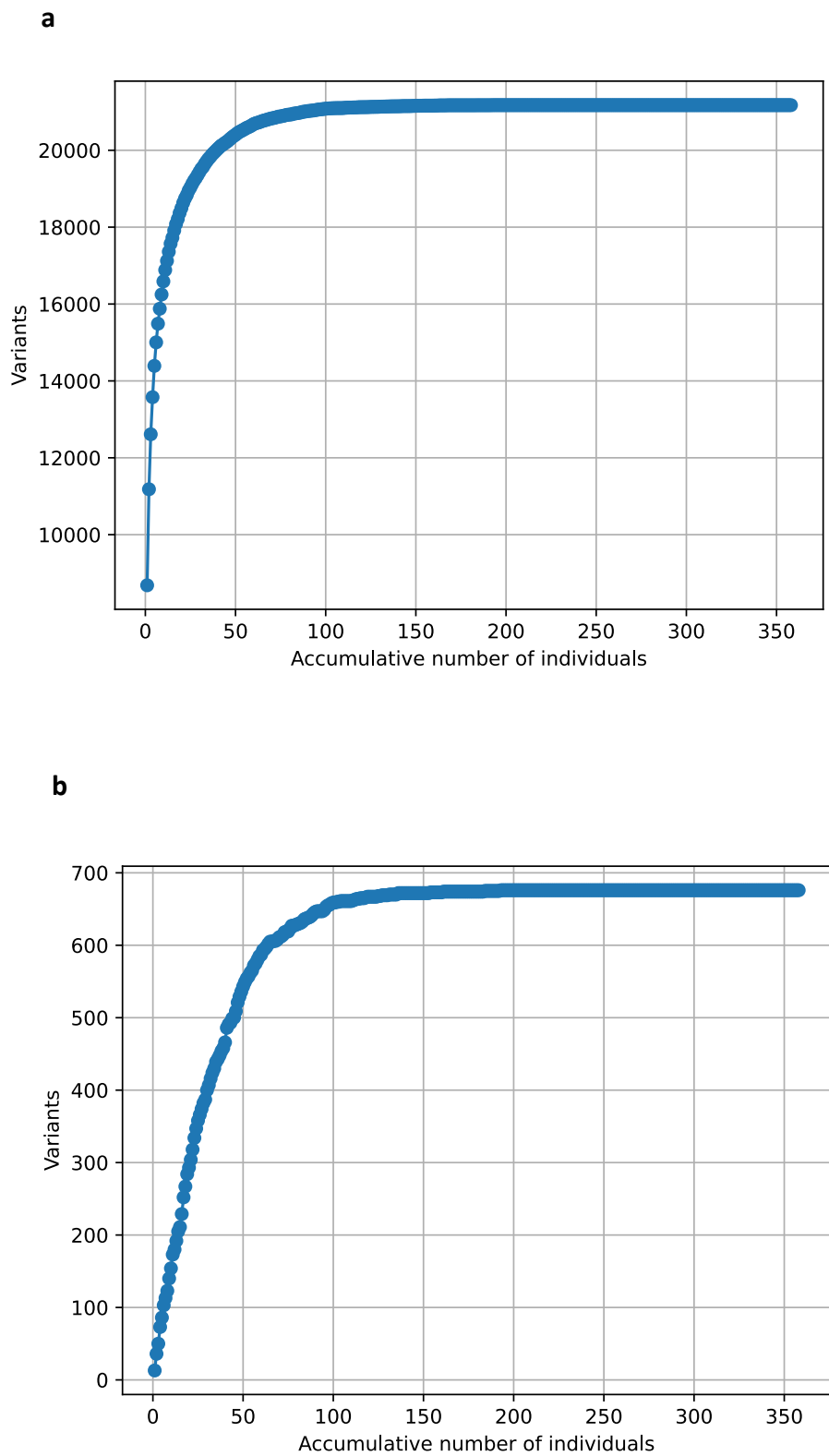
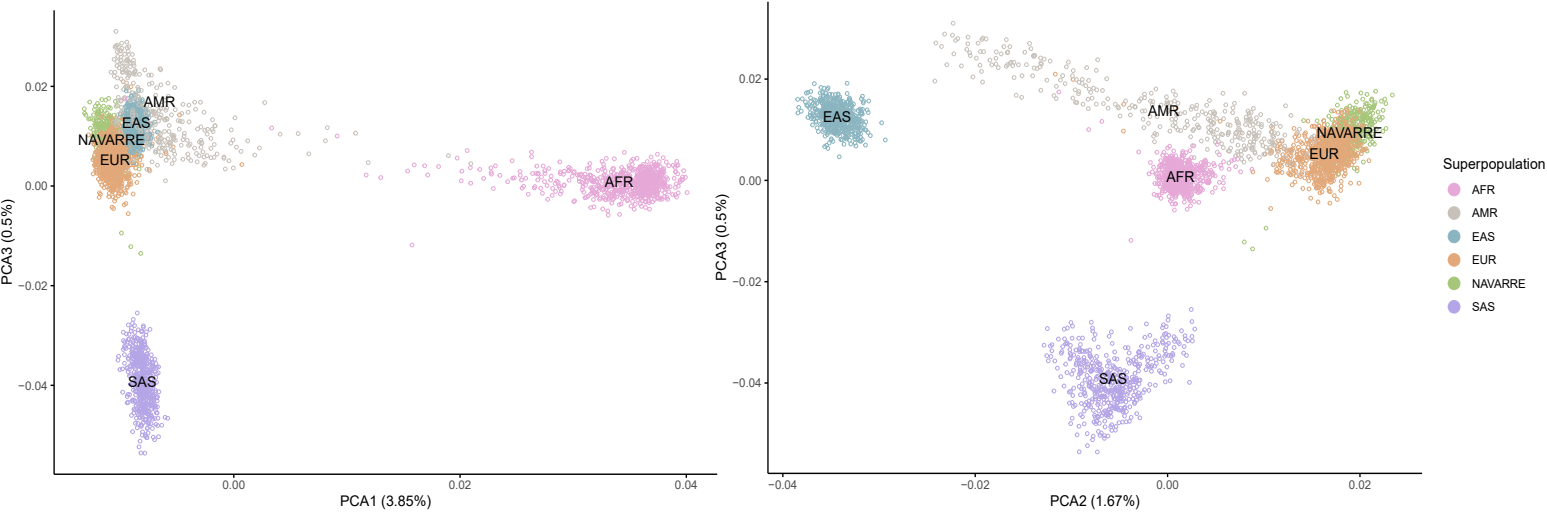


Figure S2

a



b

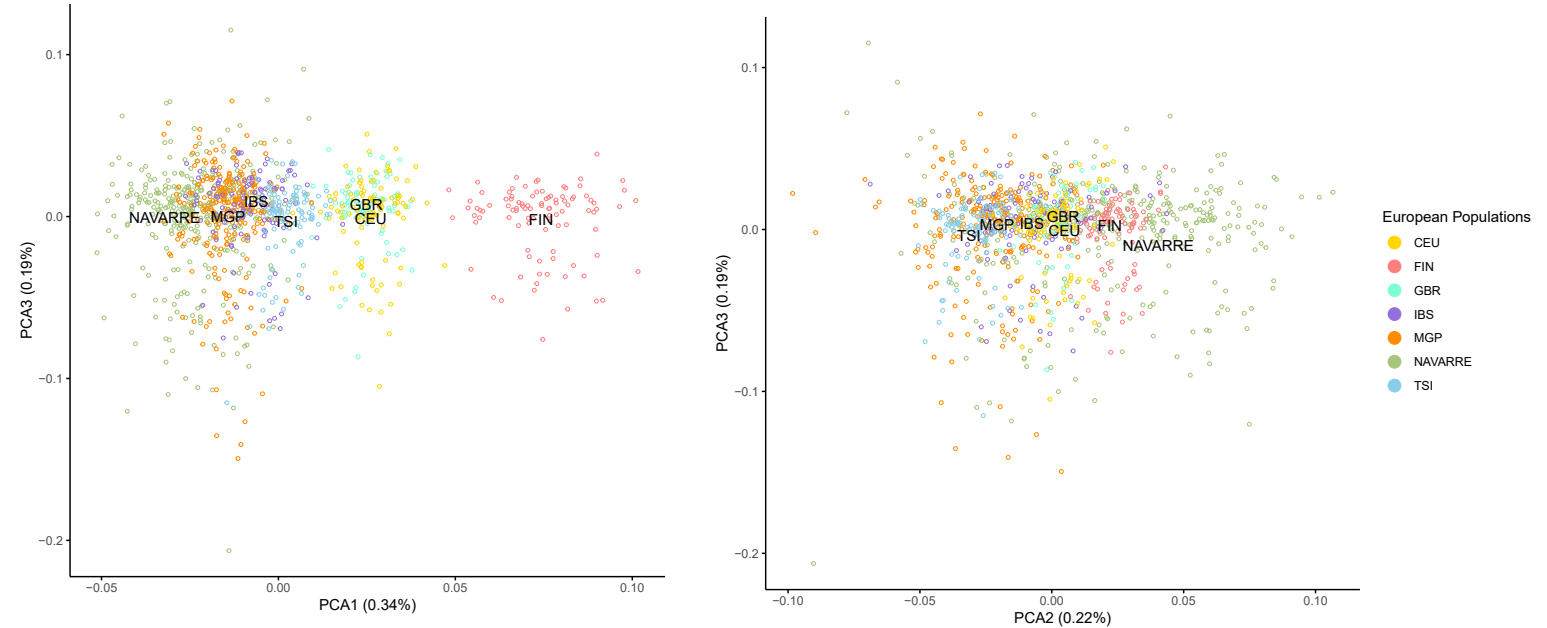


Figure S3

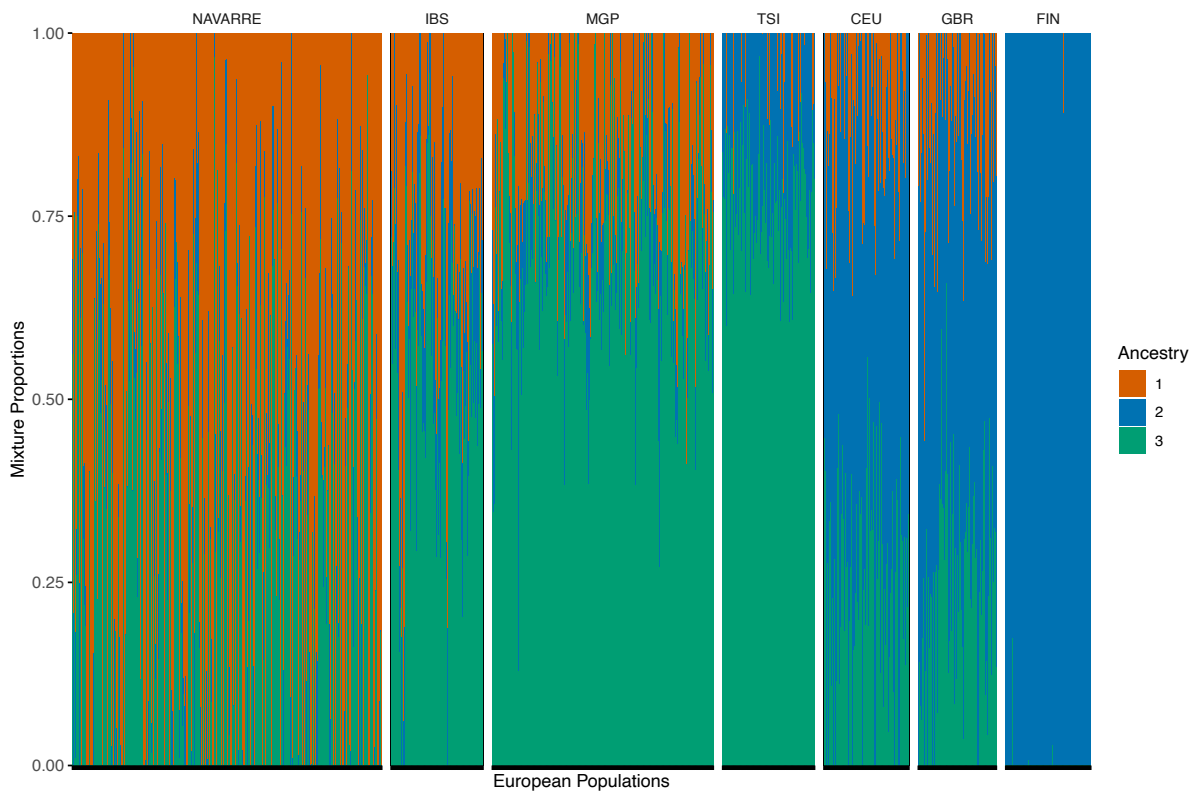


Figure S4

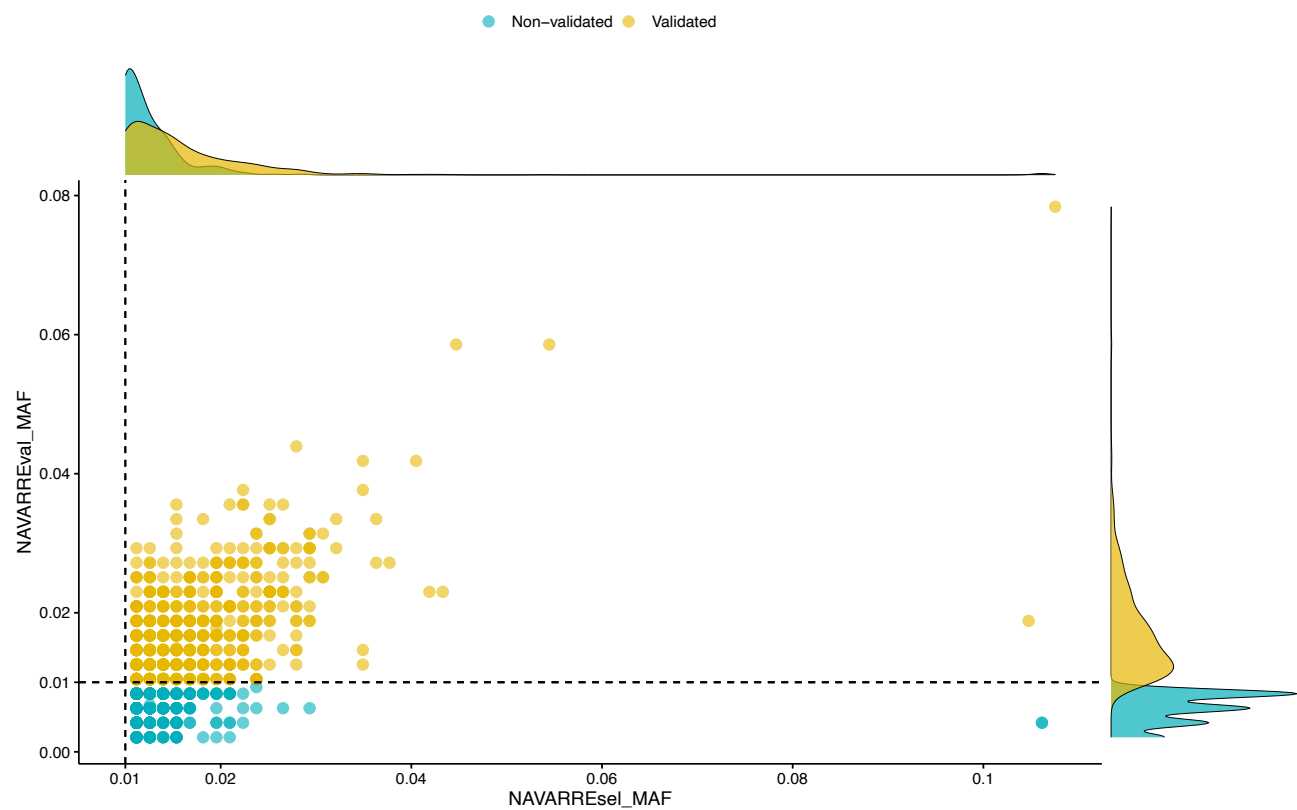


Figure S5

