

Figure S1: Extended Pedigree Configuration for Simulation Studies

The pedigree configuration for each of the 10 outbred, four-generation pedigrees included in Relationship Configuration I of the simulation studies, where the overall structure of each pedigree is as depicted, but the pattern of ancestry admixture varies according to the specified population structure setting.

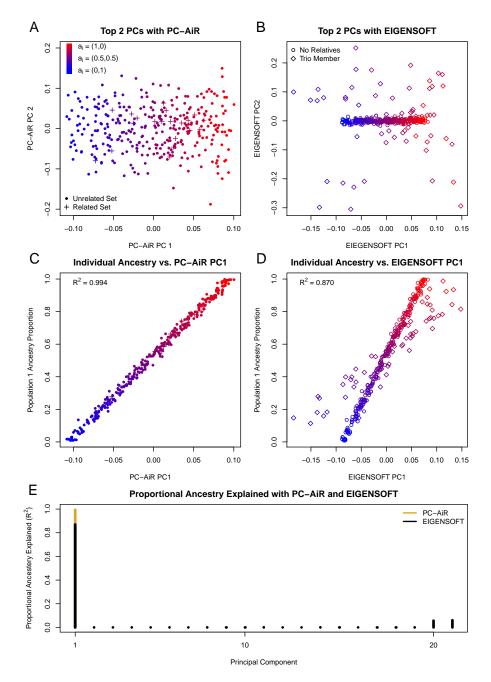


Figure S2: Comparison of PC-AiR and EIGENSOFT for Relationship Configuration II and Population Structure I with $F_{ST} = 0.01$.

(A and B) Scatter plots of principal components 1 and 2 from PC-AiR (A) and EIGENSOFT (B), respectively. (C and D) Scatter plots of the simulated population 1 ancestry proportions vs. coordinates along principal component 1 for each individual from PC-AiR (C) and EIGENSOFT (D), respectively. (A-D) The color of each point represents that individual's true ancestry; red for population 1, blue for population 2, and an intermediate color for an admixed individual. (A and C) A dot represents an individual in the ancestry representative, mutually unrelated set, and a plus represents an individual in the related set. (B and D) A diamond represents an individual. (E) Barplot of the efficiency of PC-AiR and EIGENSOFT. Each bar represents the proportion of ancestry explained (R^2 value) by each principal component from PC-AiR (gold) and EIGENSOFT (black), until a cumulative R^2 of 0.99 is achieved.

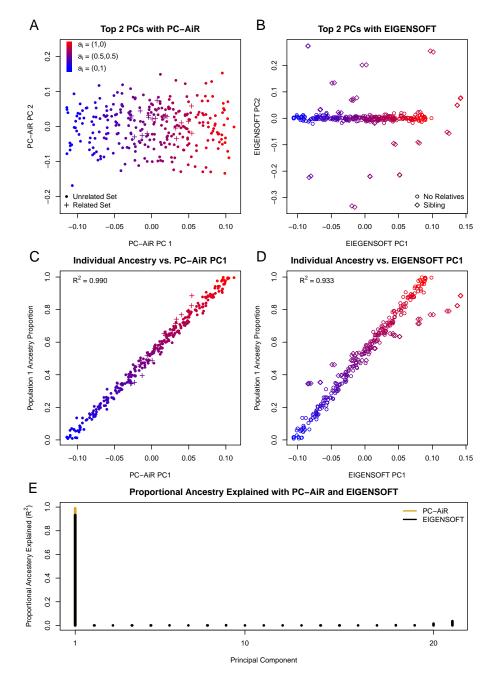


Figure S3: Comparison of PC-AiR and EIGENSOFT for Relationship Configuration III and Population Structure I with $F_{ST} = 0.01$.

(A and B) Scatter plots of principal components 1 and 2 from PC-AiR (A) and EIGENSOFT (B), respectively. (C and D) Scatter plots of the simulated population 1 ancestry proportions vs. coordinates along principal component 1 for each individual from PC-AiR (C) and EIGENSOFT (D), respectively. (A-D) The color of each point represents that individual's true ancestry; red for population 1, blue for population 2, and an intermediate color for an admixed individual. (A and C) A dot represents an individual in the ancestry representative, mutually unrelated set, and a plus represents an individual in the related set. (B and D) A diamond represents an individual who is a member of a sibling pair, and a circle represents the remaining individuals. (E) Barplot of the efficiency of PC-AiR and EIGENSOFT. Each bar represents the proportion of ancestry explained (R^2 value) by each principal component from PC-AiR (gold) and EIGENSOFT (black), until a cumulative R^2 of 0.99 is achieved.

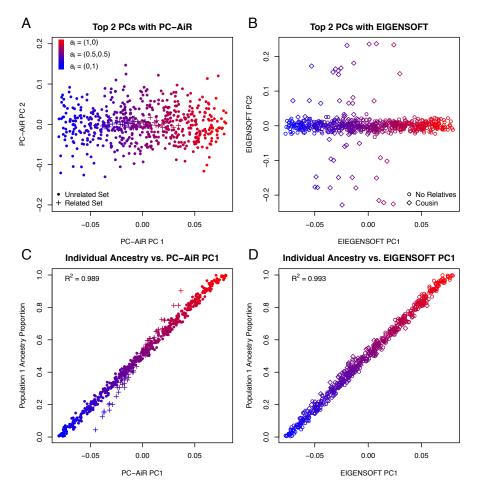


Figure S4: Comparison of PC-AiR and EIGENSOFT for Relationship Configuration IV and Population Structure I with $F_{ST} = 0.01$.

(A and B) Scatter plots of principal components 1 and 2 from PC-AiR (A) and EIGENSOFT (B), respectively. (C and D) Scatter plots of the simulated population 1 ancestry proportions vs. coordinates along principal component 1 for each individual from PC-AiR (C) and EIGENSOFT (D), respectively. (A-D) The color of each point represents that individual's true ancestry; red for population 1, blue for population 2, and an intermediate color for an admixed individual. (A and C) A dot represents an individual in the ancestry representative, mutually unrelated set, and a plus represents an individual in the related set. (B and D) A diamond represents an individual who is a member of a sibling pair, and a circle represents the remaining individuals.

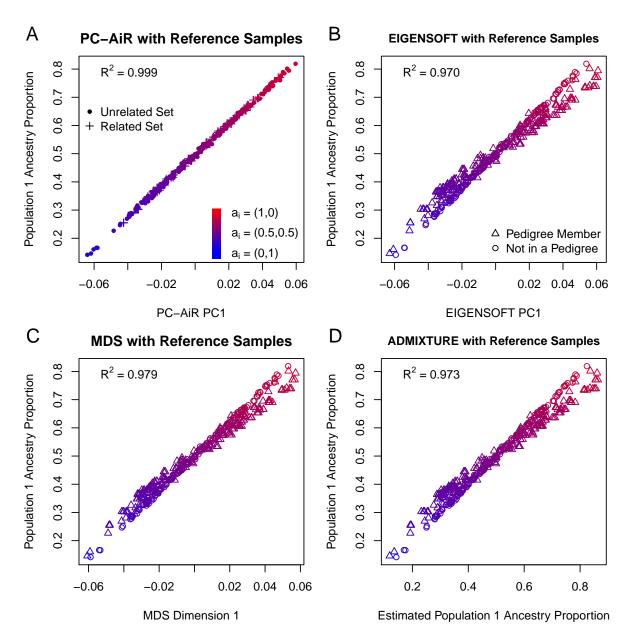
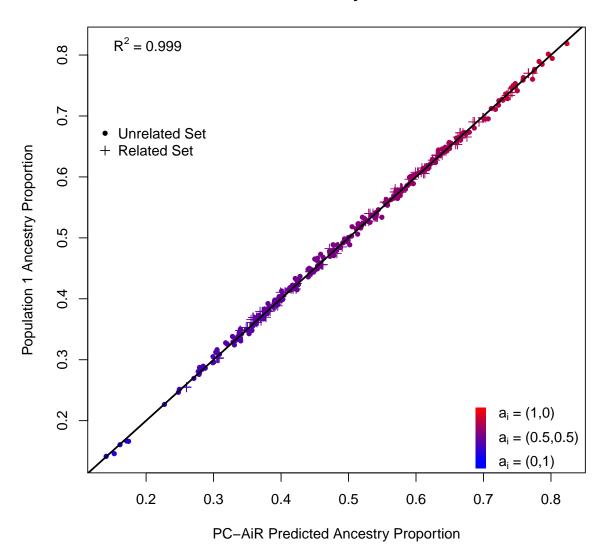
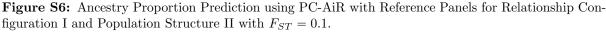


Figure S5: Population Structure Inference Results including Reference Panels for Relationship Configuration I and Population Structure II with $F_{ST} = 0.1$.

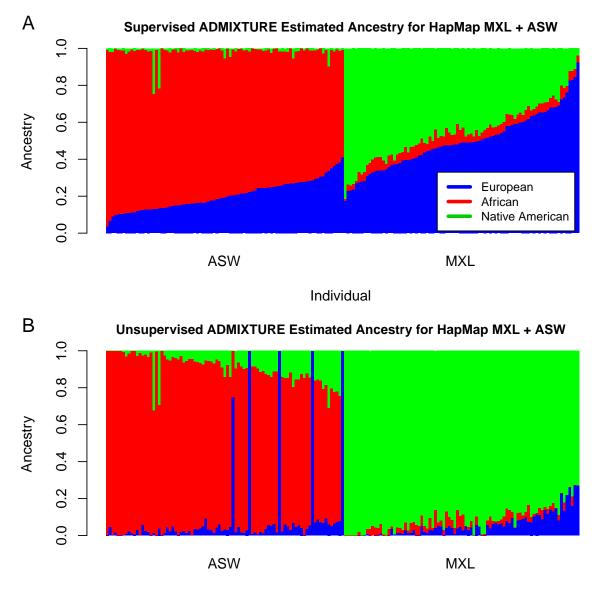
Scatter plots of the simulated population 1 ancestry proportions for each individual are plotted against: (A) coordinates along principal component 1 from PC-AiR, (B) coordinates along principal component 1 from EIGENSOFT, (C) coordinates along dimension 1 from MDS, and (D) the estimated ancestry proportions from ADMIXTURE for the inferred population with the highest R^2 . The color of each point represents that individual's true ancestry; red for population 1, blue for population 2, and an intermediate color for an admixed individual. (A) A dot represents an individual in the ancestry representative, mutually unrelated set, and a plus represents an individual in the related set. (B-D) A circle represents an individual not in a pedigree, and a triangle represents an individual who is a member of a pedigree.



PC-AiR Ancestry Prediction



Scatter plot of the simulated population 1 ancestry proportions for each individual against predicted population 1 ancestry proportions using the top principal component from PC-AiR and the methodology given by Chen et al. [2013]. The color of each point represents that individual's true ancestry; red for population 1, blue for population 2, and an intermediate color for an admixed individual. A dot represents an individual in the ancestry representative, mutually unrelated set, and a plus represents an individual in the related set.



Individual

Figure S7: HapMap MXL and ASW Individual Ancestry Bar Plots Individual ancestry estimates for 87 HapMap ASW and 86 HapMap MXL samples. (A) Supervised individual ancestry analysis with ADMIXTURE including reference population panels. Each individual is represented by a vertical bar; estimated European (HapMap CEU), African (HapMap YRI), and Native American (HGDP samples from the Americas) ancestry proportions are shown in blue, red, and green, respectively. (B) Unsupervised ancestry analysis with ADMXITURE without reference population panels. The three colors represent the three inferred ancestral populations.

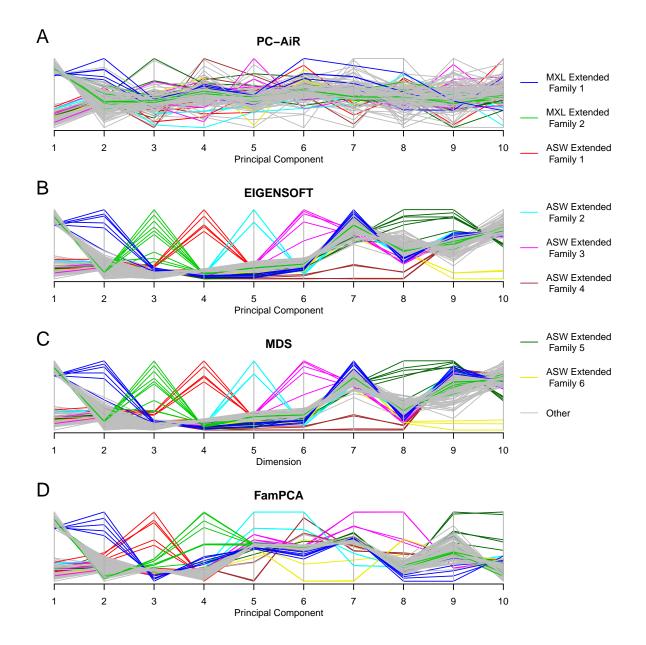


Figure S8: Parallel Coordinates Plots for the HapMap MXL and ASW Combined Sample Parallel coordinates plots of the top ten axes of variation from PC-AiR (A), EIGENSOFT (B), MDS (C), and FamPCA (D). Each vertical bar represents one of the axes of variation, and each line traces out the coordinates for an individual across all ten axes of variation. Colors are used to show individuals belonging to the same extended family. Many of the top ten axes of variation from all methods except PC-AiR are driven by the correlation of members in extended families.

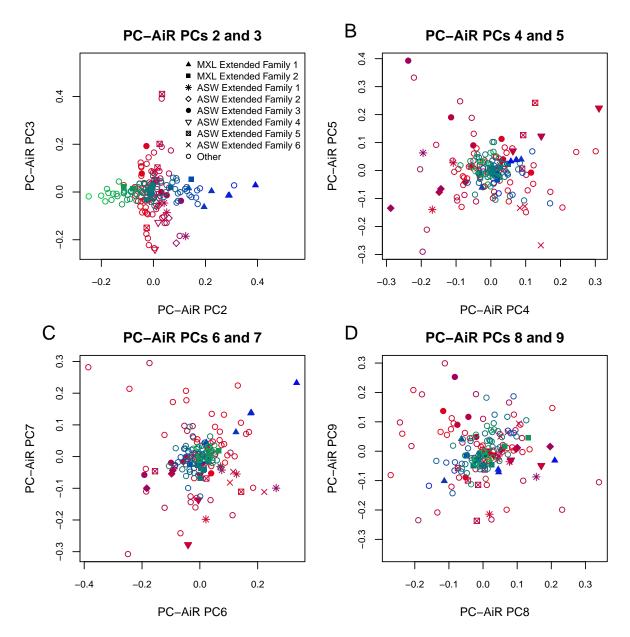


Figure S9: PC-AiR PCs 2-9 from HapMap MXL and ASW Combined Sample Scatterplots of principal components 2-9 from PC-AiR. The color of each point represents that individual's ancestry as estimated from a supervised individual ancestry analysis with ADMIXTURE; blue for European (HapMap CEU), red for African (HapMap YRI), green for Native American (HGDP samples from the Americas), and an intermediated color for an admixed individual. Different plotting characters represent individuals belonging to different extended pedigrees. PC 2 reflects European vs. Native American ancestry, but PCs 3-9 do not reflect any apparent structure.

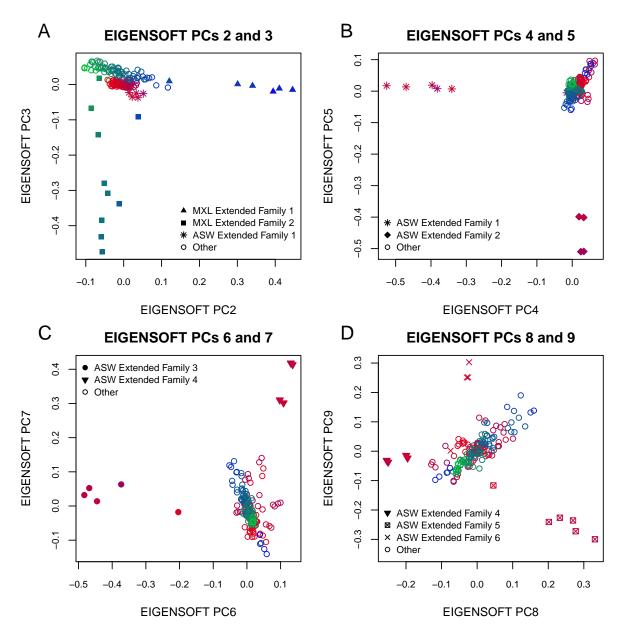


Figure S10: EIGENSOFT PCs 2-9 from HapMap MXL and ASW Combined Sample Scatterplots of principal components 2-9 from EIGENSOFT. The color of each point represents that individual's ancestry as estimated from a supervised individual ancestry analysis with ADMIXTURE; blue for European (HapMap CEU), red for African (HapMap YRI), green for Native American (HGDP samples from the Americas), and an intermediated color for an admixed individual. Different plotting characters represent individuals belonging to different extended pedigrees. All axes of variation are strongly influenced by the correlation structure of groups of relatives belonging to extended families.

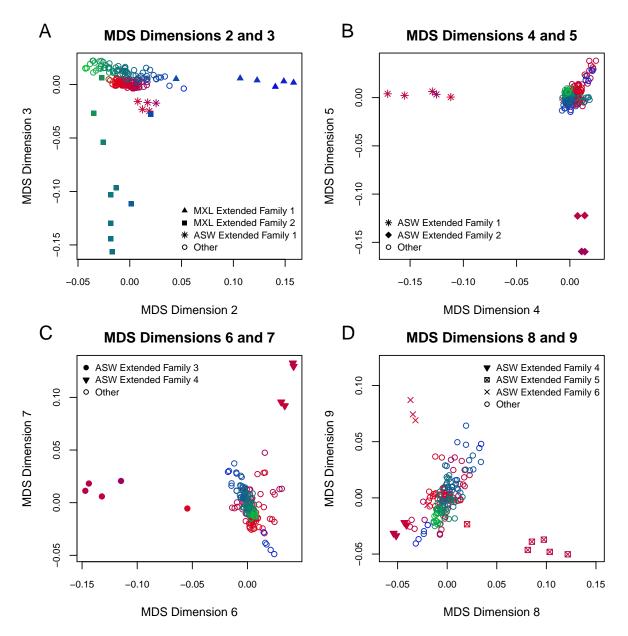


Figure S11: MDS Dimensions 2-9 from HapMap MXL and ASW Combined Sample Scatterplots of dimensions 2-9 from MDS. The color of each point represents that individual's ancestry as estimated from a supervised individual ancestry analysis with ADMIXTURE; blue for European (HapMap CEU), red for African (HapMap YRI), green for Native American (HGDP samples from the Americas), and an intermediated color for an admixed individual. Different plotting characters represent individuals belonging to different extended pedigrees. All axes of variation are strongly influenced by the correlation structure of groups of relatives belonging to extended families.

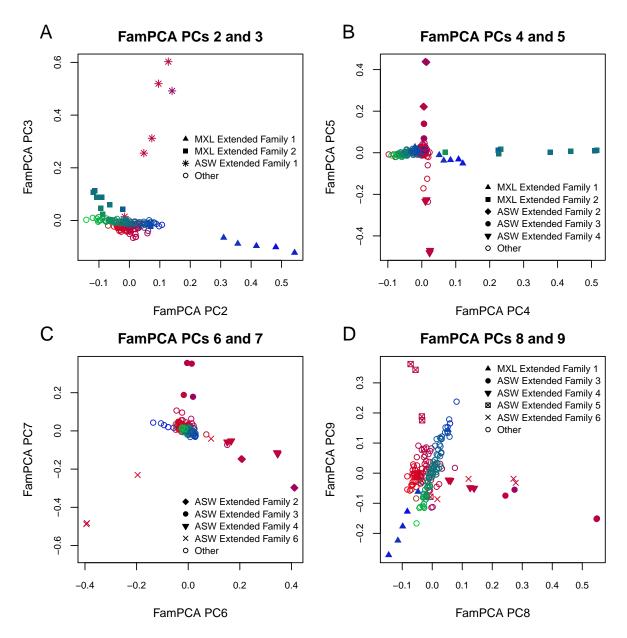


Figure S12: FamPCA PCs 2-9 from HapMap MXL and ASW Combined Sample Scatterplots of PCs 2-9 from FamPCA. The color of each point represents that individual's ancestry as estimated from a supervised individual ancestry analysis with ADMIXTURE; blue for European (HapMap CEU), red for African (HapMap YRI), green for Native American (HGDP samples from the Americas), and an intermediated color for an admixed individual. Different plotting characters represent individuals belonging to different extended pedigrees. All axes of variation are strongly influenced by the correlation structure of groups of relatives belonging to extended families.