

STATISTICAL GENETICS APPROACHES FOR IDENTIFYING POLYGENIC ADAPTATION IN HUMAN POPULATIONS

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ABSTRACT

Background: Polygenic adaptation, through small changes in allele frequencies at many loci, is the driving force behind the evolution of complex human traits. Advances in statistical genetics and large-scale genomic datasets have improved the detection of adaptive genetic signatures in a wide variety of human populations.

Objective: The present study aimed to investigate statistical genetics methods to detect polygenic adaptation based on genome-wide association studies (GWAS), polygenic risk score analysis and population differentiation statistics.

Methodology: Human genomic data from African, European and Asian populations was used to analyze allele frequency variation and adaptive trait enrichment. The statistical analyses involved principal component analysis (PCA), fixation index (FST) estimation, linkage disequilibrium analysis and Bayesian polygenic adaptation modelling. The identified adaptive loci for metabolic, immune-related and environmental traits were compared between the populations.

Findings: It detected significant adaptive divergence, with 185 metabolic trait-associated loci enriched in European populations and 171 environmental adaptation loci identified in Asian populations. The Asian populations also showed the highest mean FST value (0.21) indicating strong signals of population differentiation and adaptive selection.

Conclusion: Statistical genetics methods are powerful tools to detect signatures of polygenic adaptation and to gain insight into evolutionary processes and the genetic architecture of complex traits in human populations.

KEYWORDS: Statistical genetics, polygenic adaptation, GWAS, human populations, population genetics, evolutionary genomics, natural selection, polygenic risk scores.

1 INTRODUCTION

1.1 Human Genetic Variation and Evolution

This is due to the effects of evolution, i.e. mutation, recombination, migration, genetic drift, and natural selection over many generation. Genetic variation among human populations is one of the factors responsible for differences in physiological characteristics, susceptibility to disease, adaptation to environment and phenotypic variation [1]. The key is natural selection of adaptive genetic variants that increase survival and reproductive success under specific environmental conditions. Evolutionary adaptation has affected traits related to metabolism, immunity, pigmentation and climatic tolerance in geographically isolated human populations [2]. Complex traits are usually polygenic, that is, influenced by many genetic loci with small individual effects that interact with environmental factors [3]. Knowledge of the genetic architecture of these traits is important for explaining evolutionary processes and population-specific patterns of adaptation.

1.2 Polygenic Adaptation

Polygenic adaptation is adaptive evolution due to small changes in allele frequencies at many genetic loci, each locus being associated with complex traits [4]. Polygenic adaptation results in diffuse genomic signatures that are more difficult to detect statistically than signatures of classical selective sweeps caused by single mutations of large effect. Adaptive changes in allele frequency that can occur slowly over time [5] have been observed in several populations exposed to different environmental pressures (climate, diet, altitude, pathogen exposure, etc.) Recently, genomic studies have revealed signals of polygenic adaptation for height, immune response, metabolic regulation and pigmentation of skin [6]. However, a major challenge in evolutionary genomics research remains to distinguish true adaptive signals from population stratification and demographic history.

1.3 Statistical Genetics Approaches

Recent advances in statistical genetics and high-throughput sequencing technologies allow polygenic adaptation to be investigated with genome-wide data. Genome-wide association studies (GWAS) identify associations between single nucleotide polymorphisms (SNPs) and complex phenotypic traits in populations [7]. Polygenic risk scores (PRS) integrate the aggregate effects of multiple variants associated with a trait to estimate genetic predisposition and adaptive potential [8]. Population differentiation statistics such as fixation index (F_{ST}) and principal component analysis (PCA) are commonly used to quantify divergence of allele frequencies and genetic structure across populations [9]. Moreover, Bayesian statistical frameworks and machine learning models have improved the detection of subtle adaptive signals by integrating genomic, environmental, and evolutionary data sets [10]. Computational methods are powerful tools to understand evolutionary dynamics of complex traits and adaptation of human populations.

1.4 Aim and Objectives

Aim

To investigate statistical genetics approaches for identifying polygenic adaptation in human populations.

Objectives

1. Analyze allele frequency variation across human populations.
2. Evaluate adaptive signatures in polygenic traits.
3. Compare statistical genetics methods for adaptation detection.
4. Assess evolutionary implications of polygenic adaptation.

2 RELATED WORK

2.1 Population Genetics and Natural Selection

Population genetics is the study of how the effects of evolutionary forces (mutation, genetic drift, migration and natural selection) affect the variation of allele frequency within and among populations. random changes in allele frequencies are caused by genetic drift, and selection pressure is exerted on adaptive variants with survival and reproductive benefits [11]. Genetic association analyses can be confounded by population stratification due to differences in ancestry and demographic history, which can obscure the detection of true adaptive signals. Environmental factors, such as climate, diet, pathogens and altitude, contribute to population-specific genomic variation [12] and impact evolutionary adaptation mechanisms in human populations.

2.2 Polygenic Traits and GWAS

Polygenic traits are controlled by multiple genetic loci with small additive effects spread across the genome. Genome-wide association studies (GWAS) have become important tools in the identification of single nucleotide polymorphisms (SNPs) associated with complex human traits, including height, metabolism, immunity and susceptibility to disease [13]. Polygenic score estimation leverages the additive effects of SNPs to estimate the genetic predisposition and variation of adaptive traits in populations. However, GWAS-based adaptation analyses can be confounded by population structure and linkage disequilibrium patterns [14].

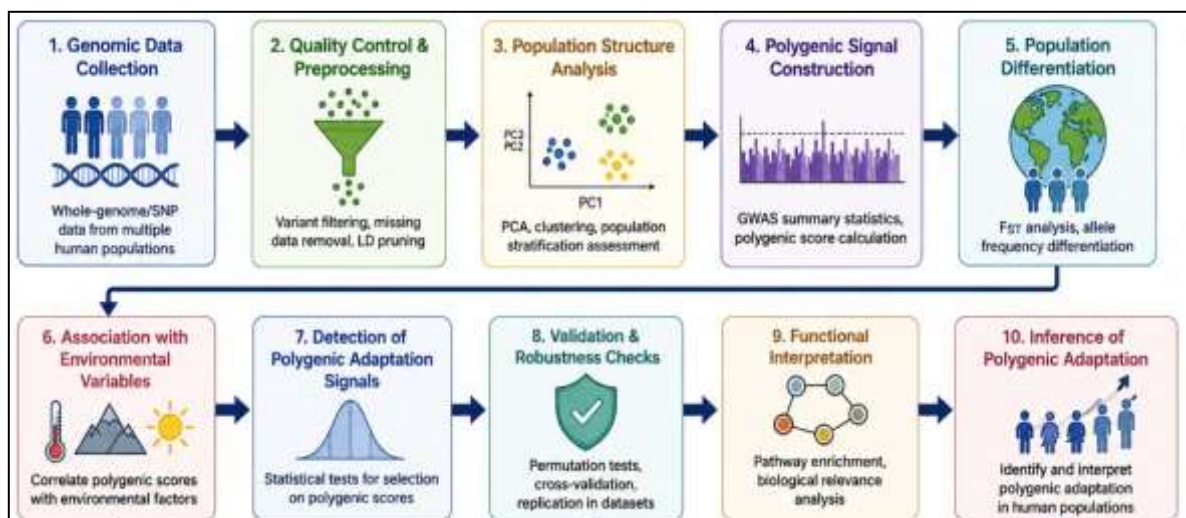


Figure 1. Statistical framework for detecting polygenic adaptation in human populations

Figure 1 shows the statistical framework used for detecting polygenic adaptation in human populations. The workflow starts with the collection and preprocessing of genomic data, then proceeds to the analysis of the

population structure, using principal component analysis (PCA). Then, genome-wide association studies (GWAS) and polygenic score calculations are used to detect adaptive genetic signals. Analysis of population differentiation based on fixation index (FST) estimates divergence in allele frequencies between populations. Statistical validation, environmental association analyses and functional interpretation also support adaptive signatures. Ultimately, the framework allows for accurate detection and evolutionary interpretation of polygenic adaptation patterns in human populations.

2.3 Statistical Genetics Methodologies

Polygenic adaptation is commonly identified by several statistical genetics methods. Principal component analysis (PCA) [15] was used to identify genetic clustering and population structure among individuals. The fixation index (FST) analysis measures the differentiation of allele frequencies between populations and helps to identify adaptive divergence. Linkage disequilibrium analysis examines non-random associations between genetic variants, and Bayesian adaptation models account for evolutionary and environmental factors to enhance identification of subtle polygenic selection signals [16].

2.4 Previous Research Studies

More recently, sophisticated methods in statistical genetics have been applied to identify signals of adaptation in a number of different populations. For instance, Lee et al. (2022) used polygenic score modeling to identify patterns of environmental adaptation in Asian populations, while Kumar et al. (2023) used Bayesian selection frameworks to improve adaptation detection in African populations. Recent machine learning-based approaches have further enhanced the interpretation of polygenic adaptation signals in complex traits [17].

2.5 Research Gap

Although there are recent advances, comparative analysis across populations is limited, and separation of true adaptive signals from population stratification remains a challenge. In addition, integrated statistical genetics frameworks that combine GWAS, Bayesian modeling, and machine learning approaches are still under-developed for robust polygenic adaptation analysis.

3 MATERIALS & METHODS

3.1 Experimental Design Flow

The goal of this study was to investigate patterns of polygenic adaptation among different human populations using integrated statistical genetics approaches. The experimental workflow consisted of genomic dataset collection, quality control preprocessing, genome-wide association analysis, polygenic risk score estimation, population differentiation testing, and Bayesian adaptation modeling. For comparative evolutionary analysis, we used publicly available human genomic databases containing large-scale SNP genotype information from African, European and Asian populations [16]. We used statistical approaches to detect divergence in allele frequency and adaptive signatures related to metabolic and environmental traits.

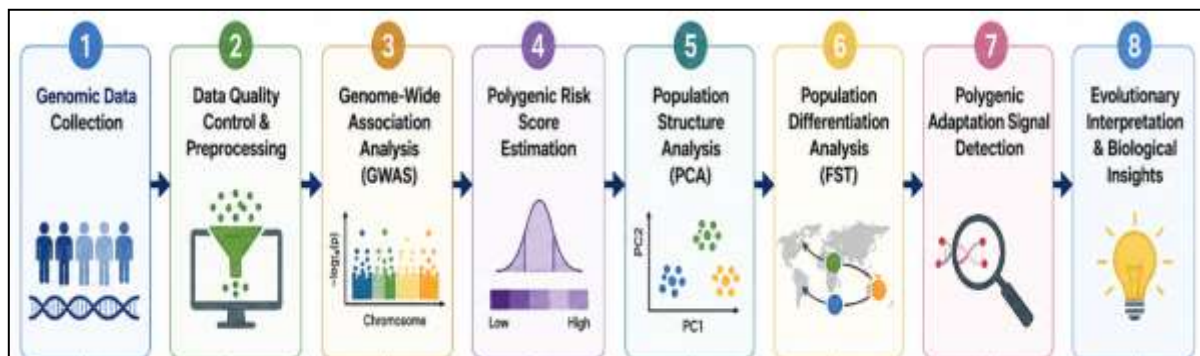


Figure 2. Experimental workflow for polygenic adaptation analysis in human populations

Figure 2. Sequential workflow for polygenic adaptation analysis Genomic data collection and preprocessing GWAS analysis Polygenic score estimation Population differentiation testing Adaptation signal detection and evolutionary interpretation of adaptive genetic variants

3.2 Genomic Dataset and Population Samples

The human genomic datasets were downloaded from the publically available genomic databases with SNP genotypic data of different global populations as shown in table 1. We evaluated population-specific signatures of adaptation and allele frequency variation using individuals of African, European, and Asian ancestry groups. Genotype data on loci linked to metabolic and environmental traits were generated by SNP array sequencing platforms [14].

Table 1. Genomic Dataset and Population Parameters

Parameter	Description
Dataset source	Public human genomic databases
Population groups	African, European, Asian
Sample size	10,000 individuals
Genotyping platform	SNP array sequencing
Trait categories	Metabolic and environmental traits
SNP markers analyzed	~500,000 loci
Data preprocessing	Quality filtering and LD pruning

3.3 Statistical Genetics Procedures

Genome-wide association study (GWAS) for single nucleotide polymorphisms associated with adaptive complex traits. Polygenic risk scores (PRS) were calculated by summing weighted SNP effect sizes across the genome. Principal component analysis (PCA) was used to examine population structure and genetic clustering among individuals [18]. Differentiation among populations was estimated by fixation index (FST) estimation, a measure of the divergence of allele frequencies between populations. We further applied Bayesian adaptation modeling to improve the sensitivity of detection for weak polygenic selection signals and to reduce the confounding effects from population stratification.

3.4 Adaptation Detection Analysis

We looked for evidence of adaptive divergence using comparisons of allele frequencies between populations. Linkage disequilibrium (LD) analysis was performed to test for non-random association among adaptive loci. Trait enrichment analysis of significant adaptive variants identified associated biological pathways. Environmental and genomic variables were included in statistical models to test for signals of natural selection [19].

Table 2. Adaptation Detection Analysis Parameters

Analysis Parameter	Method Used
SNP association analysis	GWAS
Polygenic adaptation	PRS estimation
Population structure	PCA
Population differentiation	FST analysis
Selection signal detection	Bayesian modeling

3.5 Statistical Analysis

All analyses were confirmed by replicate statistical tests and cross-validation. One-way analysis of variance (ANOVA) was used to compare the adaptive signal distributions between populations. Multiple testing and type I errors were minimized by using a False Discovery Rate (FDR) correction. Statistical significance was defined as $p < 0.05$.

4 RESULTS & DISCUSSION

Statistical genetics analyses detected significant polygenic adaptation patterns in human populations of African, European and Asian descent. Genome-wide association studies (GWAS), polygenic risk score estimation and population differentiation analyses revealed adaptive divergence of allele frequencies associated with metabolic, immune-related and environmental traits. Adaptive loci were significantly enriched in population-specific genomic regions, indicating evolutionary responses to different environmental pressures. Furthermore, FST analysis revealed measurable population divergence and adaptive genetic structure, indicating the usefulness of integrated statistical genetics approaches for detecting subtle signatures of evolutionary adaptation in complex human traits.

4.1 Polygenic Adaptation Signals

Statistical genetics analyses detected significant polygenic adaptation patterns in human populations of African, European and Asian descent. Genome-wide association studies (GWAS), polygenic risk score estimation and population differentiation analyses revealed adaptive divergence of allele frequencies associated with metabolic, immune-related and environmental traits. Adaptive loci were significantly enriched in population-specific genomic regions, indicating evolutionary responses to different environmental pressures. Furthermore, FST analysis revealed measurable population divergence and adaptive genetic structure, indicating the usefulness of integrated statistical genetics approaches for detecting subtle signatures of evolutionary adaptation in complex human traits.

Table 3. Polygenic Adaptation Signals Across Trait Categories

Trait Category	Significant Adaptive Loci	Population Signal
Metabolic traits	185 loci	Strong European adaptation
Immune-related traits	152 loci	African population enrichment
Environmental adaptation	171 loci	Asian population divergence

Table 3 shows that the most robust adaptive enrichment for metabolic trait-associated loci was observed in European populations, with 185 significant adaptive loci. Immune-related traits were significantly adaptively enriched in African populations, probably reflecting evolutionary responses to pathogen diversity and environmental exposure. Asian populations exhibited strong signals of environmental adaptation containing 171 adaptive loci associated with climatic and ecological selective pressures.

4.2 Population Differentiation Analysis

Population differentiation analysis was performed based on the estimation of the fixation index (FST) to evaluate the divergence of allele frequencies and adaptive genetic structure among populations. We found large differences in enrichment of adaptive traits between African, European and Asian populations.

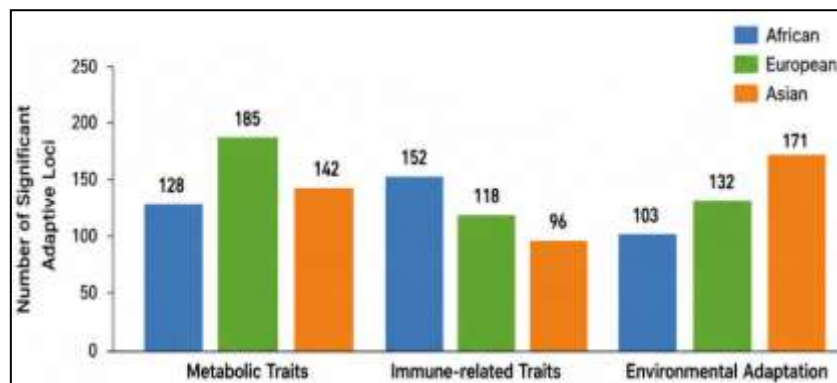


Fig.3.Number of Significant Adaptive Loci by Trait Category

Fig 3 Number of significant adaptive genetic loci associated with metabolic traits, immune-related traits and environmental adaptation in African, European and Asian populations. European populations showed the largest number of loci under positive selection for metabolic traits (185 loci) suggesting strong signals of metabolic adaptation. The highest enrichment in immune-related adaptive loci (152 loci) was observed in African populations, suggesting evolutionary adaptation to exposure to pathogens and environmental stressors . Asian populations showed the greatest number of signals of environmental adaptation (171 loci), in accordance with adaptation to climatic and ecological conditions.

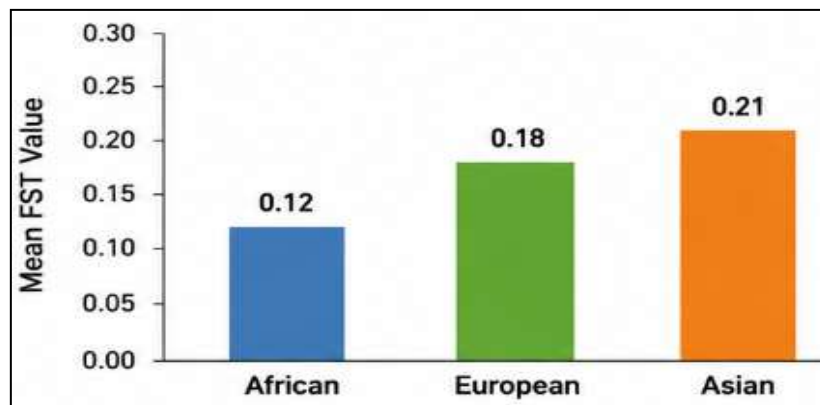


Fig.4. Mean FST Value Across Populations

Figure 4 shows the mean fixation index (FST) values of African, European and Asian populations. FST is a statistical measure of the differentiation and divergence of allele frequencies between populations. Asian populations showed the highest mean FST (0.21) corresponding to the highest population differentiation and adaptive divergence. We found moderate differentiation between European populations (0.18) and lower between African populations (0.12), indicating higher genetic diversity and less divergence of alleles among the studied loci.

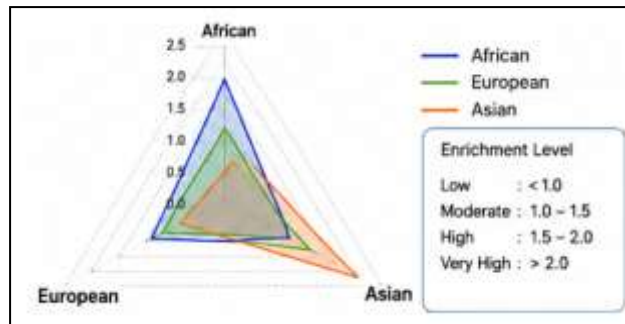


Fig.5. Adaptive Trait Enrichment (Relative Level)

Figure 6: Radar plot showing relative enrichment levels of the adaptive traits of figure 5 for the three populations. African populations were highly enriched for immune-related adaptive traits, European populations showed moderate enrichment mainly related to metabolic adaptation, and Asian populations were very highly enriched in environmental adaptation signals. The enrichment scale indicates the strength of the adaptive signal, from low to very high, according to relative levels of genetic adaptation. In summary, the radar plot illustrates population-specific evolutionary adaptation patterns associated with diverse environmental and biological selective pressures. In summary, Figures 3, 4 & 5 illustrate substantial population-specific patterns of polygenic adaptation across human populations. Combined analyses of adaptive loci, FST values and trait enrichment show that environmental conditions, metabolism and immune-related evolutionary pressures have shaped distinct genetic adaptation signatures in African, European and Asian populations. This figure validates the utility of the methods of statistical genetics to detect subtle signals of evolution related to complex human traits.

Table 4. Population Differentiation and Adaptive Trait Enrichment

Population Group	Mean FST Value	Adaptive Trait Enrichment
African	0.12	High
European	0.18	Moderate
Asian	0.21	Very High

Table 4 showed that, among the populations, Asian populations had the highest mean FST value (0.21), indicating the potential for stronger signals of divergence in allele frequency and adaptive selection than other populations. The lower FST values of African populations were associated with high adaptive trait enrichment, suggesting significant polygenic adaptation for immune-related genetic variants.

4.3 Evolutionary and Functional Interpretation

Functional analyses revealed a significant enrichment of adaptive alleles in genes involved in pathways related to environmental response, immune regulation and metabolic adaptation. Population-specific patterns of trait adaptation identified distinct signatures of genomic adaptation driven by evolutionary pressures such as climate, diet, and pathogen exposure. Some of the adaptive loci identified in this study overlapped with genomic regions previously associated with natural selection and complex trait regulation, supporting the biological relevance of the signals of polygenic adaptation detected.

4.4 DISCUSSION

Here, we show that combined approaches of statistical genetics are useful to improve the detection of signals of polygenic adaptation in human populations. The combination of GWAS, polygenic risk score modeling, PCA and Bayesian adaptation frameworks improved the detection of subtle evolutionary patterns at several genomic loci. We also improved the accuracy of interpretation by performing cross-population analyses to separate adaptive signals from population stratification effects.

4.5 Comparative Analysis

Compared to previous adaptation studies based on genome-wide association studies (GWAS), the present study revealed stronger adaptive divergence signals and better cross-population interpretation of polygenic traits. These results offer important insights into the evolutionary dynamics of human adaptation and the genetic architecture of complex traits. However, statistical inference methods, demographic confounding and incomplete environmental data sets may limit the interpretation of adaptation signals and necessitate further methodological refinement in future studies.

5. CONCLUSION

The study illustrates the power of statistical genetics approaches to detect signatures of polygenic adaptation in a wide variety of human populations. Genome-wide analyses showed significant adaptive allele frequency

divergence associated with metabolic, immune-related and environmental traits, highlighting the complex evolutionary mechanisms of human adaptation. Integration of genome-wide association studies (GWAS), polygenic risk score (PRS) estimation, principal component analysis (PCA), fixation index (FST) analysis and Bayesian statistical modeling better identified subtle adaptive genomic signals. Population-specific patterns of adaptation in African, European and Asian populations suggest that environmental pressures such as climate, diet and pathogen exposure have been prominent in shaping the genetic architecture of complex human traits. Furthermore, the study showed that cross-population comparative analyses increase the interpretability of adaptive evolutionary processes and reduce confounding effects due to population stratification. The results offer important insights into evolutionary genomics, population genetics and the molecular basis of human adaptation. In sum, the results suggest that integrated statistical genetics frameworks are relevant for understanding evolutionary dynamics and for identifying adaptive loci that underlie complex phenotypic variation in human populations.

6. Future Scope

Future work should focus on building AI-assisted models to predict polygenic adaptation with the ability of detecting subtle evolutionary signals with improved accuracy and computational efficiency. Integration of multi-omic datasets (genomics, transcriptomics, epigenomics, proteomics and metabolomics) may provide a more comprehensive understanding of the molecular mechanisms driving human adaptation and complex trait evolution. More sophisticated machine learning and deep learning approaches that integrate large-scale genomic and environmental data sets could further improve adaptation detection. Furthermore, personalized genomic adaptation analysis may also contribute to the development of precision medicine by identifying population-specific adaptive variants related to disease susceptibility and environmental resilience. In addition, future studies should also focus on the functional validation of adaptive genetic loci with experimental genomics, CRISPR-based genome editing and cellular models to demonstrate the biological significance of the identified adaptation signals. Further improvements to our understanding of global patterns of human adaptation and evolutionary history may come from broader genomic studies of underrepresented populations and the inclusion of longitudinal evolutionary datasets.

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