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Article

Investigating the contribution of genetic risk factors to gastric cancer in Korean-ancestry populations

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SUMMARY

Korean-ancestry populations and East Asians broadly have had historically high rates of gastric cancer. Genetic contributors to this high incidence have not been thoroughly explored in Koreans. This project investigated whether previously identified genetic risk factors in non-Korean populations were associated with high gastric cancer incidence rates in Koreans. We hypothesized that there would be higher risk allele frequencies for gastric cancer in Koreans, and that most single-nucleotide polymorphisms (SNPs) identified in other populations as being associated with gastric cancer would have concordant effects in a Korean population. Using public genome-wide association study (GWAS) data, we conducted three analyses. First, our remapping analysis identified 120 SNPs that are more closely linked in Koreans to the causal variants for gastric cancer that are shared across populations. Second, our direction of effect analysis revealed that 68.42% of significant SNPs exhibited consistent effects across South Korean and other populations. Lastly, East Asian populations in the 1000 Genomes Project did not show significantly higher average risk allele frequencies for gastric cancer-associated SNPs compared to other populations, which could indicate that environmental factors play a larger role or that key causal variants remain unidentified. While known gastric cancer SNPs implicated in non-Korean populations appear to have consistent effects in ethnically Korean populations, different tag SNPs in Korean populations may be more strongly linked to the underlying causal variants. The role of genetics in the high gastric cancer incidence observed in East Asian populations appears limited based on comparisons of risk allele frequencies across populations.

INTRODUCTION

Gastric cancer, also referred to as stomach cancer, affects the epithelial lining of the stomach, most commonly the gastric mucosa, and encompasses different anatomical subsites including the cardia and non-cardia regions. It is the fifth most common cancer and has the fourth-highest mortality rate worldwide (1, 2). While the impact of this disease is widespread, East Asia is observed to have the highest rates of gastric cancer, with the age-standardized incidence rates for gastric cancer in East Asia being around double that of the regions with the second highest incidence rate, Central Europe and Eastern Europe (3). South Korea has historically dealt with high rates of gastric cancer. In 2002, the Korean Gastric Cancer Association (KGCA) reported gastric cancer as the most common cancer in the country (4). Furthermore, South Korea currently has the third-highest incidence rate of gastric cancer in the world, only behind Mongolia and Japan: compared to the global incidence rate of 11.1 per 100,000, South Korea has a rate of 27.9 per 100,000 (2). In the United States of America (USA), ethnic Koreans disproportionately contribute to the number of stomach cancer cases (5). Gastric cancer risk in South Korea and other East Asian populations has been associated with high prevalence of *Helicobacter pylori* (*H. pylori*) infection, diet, alcohol consumption, and smoking rates (6). However, it is possible that these environmental risk factors are not the only contributors to high gastric cancer incidence rates in South Korea and other East Asian populations. Other diseases, including breast cancer, are known to be affected by genetic factors (7). As there is a precedent, East Asian populations may possess a higher genetic susceptibility for gastric cancer. Research into these genetic factors may allow for better risk prediction for gastric cancer in people of Korean ancestry.

Several genome-wide association studies (GWAS) for gastric cancer have identified single-nucleotide polymorphisms (SNPs) with genome-wide significant associations. For example, the Study Group of Millennium Genome Project for Cancer identified SNPs at *PSCA* as significantly associated with diffuse gastric cancer (8). Saeki et al. found *MUC1* to also be associated with diffuse gastric cancer susceptibility (9). Although some studies have included East Asian populations, many GWAS have been conducted in populations of other ancestries. Efforts to replicate studies for commonly known variants in South Korean populations have often yielded limited or inconsistent results (10). Furthermore, hereditary diffuse gastric cancer (HDGC), most commonly associated with *CDH1* variants, is well characterized in Western populations but remains not as thoroughly explored in East Asians, with unclear incidence and variant distribution (10). Additionally, numerous studies have identified genes that interact with environmental factors. For example, a variant at *ALDH2* common in East Asian populations may increase cancer risk by impairing the detoxification of toxic aldehydes generated during alcohol metabolism (11, 12, 13, 14). A trans-ethnic study on Asians and non-Asians indicated that around one-fifth of diffuse gastric cancers could be influenced by alcohol consumption in conjunction with inactive *ALDH2* or *CDH1* variations (15). Other variants with environmental interactions are also not well investigated among Korean populations.

We examined whether the genetic risk factors identified in previously studied populations were shared in Korean populations. We hypothesized that there would be higher risk allele frequencies for gastric cancer in Koreans, and that most

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SNPs associated with gastric cancer from other studies would be associated with gastric cancer risk in a Korean-ancestry population. We found that SNPs associated with gastric cancer based on GWAS conducted in non-Korean-ancestry populations generally shared similar effects in a Korean-ancestry cohort. We also identified SNPs more significantly associated with gastric cancer in the South Korean population based on these known GWAS associations, which might serve as GWAS tag variants more suitable for populations of Korean or East Asian ancestry. We observed that populations in East Asia did not have elevated average allele frequencies for gastric cancer-associated SNPs compared to other regions around the world.

RESULTS

Genetic factors contributing to gastric cancer risk have not been as thoroughly investigated in populations of Korean ancestry compared to other populations. To investigate the role of genetics in elevated gastric cancer rates, we hypothesized that most gastric cancer associated SNPs from GWAS in other populations would explain the high gastric cancer risk in Korean-ancestry and other East Asian-ancestry populations. We first attempted to identify SNPs with stronger associations to gastric cancer specifically in South Koreans. We then examined whether variants were transferable between South Korean and other populations through comparing the direction of effect for the SNPs. We then tested differences in risk allele frequencies (i.e., the proportion of all alleles in a population that increases the risk of some disease) to observe genetic differences in varying populations.

Remapping analysis

We first conducted a remapping analysis to identify gastric cancer-associated SNPs from the GWAS Catalog that were most significantly associated with gastric cancer in a Korean-ancestry population. We did this using GWAS data from the Korean Genome and Epidemiology Study (KoGES), a cohort of participants from South Korea. This analysis is based upon the concept of linkage disequilibrium (LD), which is a statistical measure that quantifies the association between variants such that variants closer together are more likely to be inherited together and have higher LD. Tag variants are in LD with the casual variants directly contributing to gastric cancer risk. Assuming SNPs with previous associations with gastric cancer are tag variants for shared causal variants, this analysis identifies SNPs with stronger associations to the causal variants in Korean-ancestry populations. Out of the 122 SNPs in the GWAS Catalog associated with gastric cancer and not discovered in studies of Korean-ancestry populations, 106 SNPs overlapped with the SNPs available in the KoGES GWAS. We identified 120 SNPs that had stronger associations with gastric cancer in the KoGES cohort in the vicinity of the GWAS Catalog SNPs (**Figure 1**). For example, the GWAS Catalog SNP rs12471190, located near the gene *LINC02580*, had a *p*-value of 4.4×10^{-2} in the KoGES GWAS, and through remapping, we identified a SNP 26,480 base pairs upstream (rs4245784) with a more significant *p*-value of 4×10^{-6}. It should be noted that the genome wide significance threshold for *p*-values in a GWAS is 5×10^{-8}, and no original or new SNPs reached this threshold (16). Some original SNPs have multiple corresponding new SNPs because they have the same *p*-value.

Direction of effect analysis

Given the limited statistical power of the KoGES GWAS, we examined the transferability of genetic risk variants between Korean populations and non-Korean populations, which were predominantly European in the GWAS Catalog. Specifically, we checked if the effects of the risk alleles were in the same direction more often than expected by chance by comparing the observed direction of effect given by a SNP's odds ratio (OR) or beta value, which reflects the magnitude and direction of effect on gastric cancer risk for a given variant, in the KoGES GWAS and GWAS Catalog. We found that 68.4% of SNPs genome-wide significant for gastric cancer in the GWAS Catalog exhibited directionally concordant effects in Korean populations (**Figure 2**). We conducted a binomial test showing that this proportion is significantly greater than what is expected for results under random chance (*p*-value = 4.25×10^{-4}).

Allele frequency comparison analysis with GWAS Catalog SNPs

To examine the contribution of genetics to the high rates of gastric cancer in East Asians, we compared the average difference of risk allele frequencies in the 1000 Genomes Project for SNPs in the GWAS Catalog for the five major populations: Africa (AFR), the Americas (AMR), East Asia (EAS), Europe (EUR), and South Asia (SAS) (17). If there is a genetic contribution to disparities in gastric cancer risk across populations, then we expect that the populations with relatively high rates of cancer to have higher risk allele frequencies on average. We note that Koreans were not sampled in the 1000 Genomes Project but given the relatively high gastric cancer

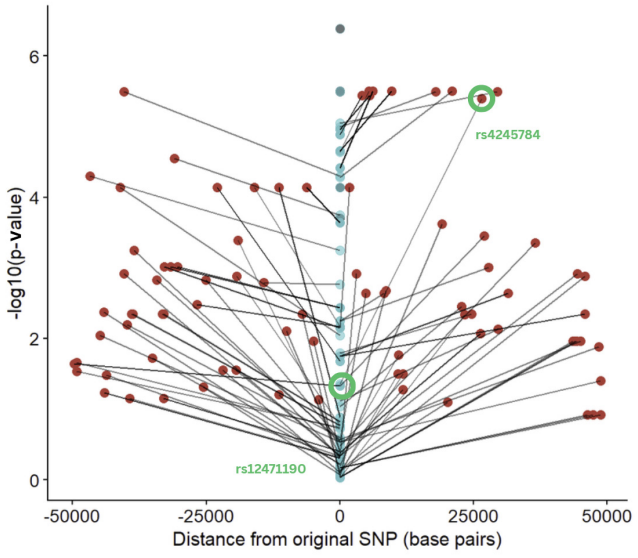

Figure 1: Newly identified tag SNP positions and p-values in the KoGES GWAS. Original SNPs (GWAS Catalog SNPs in the KoGES GWAS) are shown in blue on the vertical line at the 0 position on the x-axis. For each of these SNPs, a line is drawn connecting it to the new SNP that was identified from it, which is shown in red. Newly identified SNPs are positioned by -log10(p-value) on the y-axis and distance from the original SNP in base pairs on the x-axis. The two SNPs circled and labeled in green are referenced in the remapping analysis paragraph in the results section as an example. SNP: Single nucleotide polymorphism. GWAS: Genome-wide association study. KoGES: Korean Genome and Epidemiology Study.

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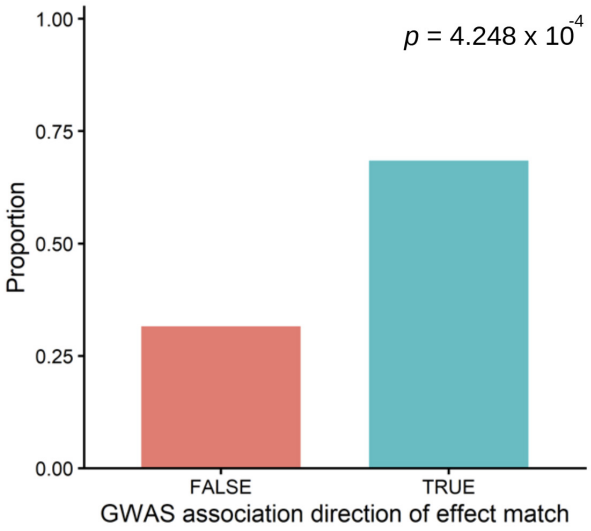

Figure 2: SNPs in KoGES GWAS with matching direction of effect as the corresponding SNPs in GWAS Catalog. Bar graph showing proportion of whether a SNP's direction of effect between the KoGES GWAS and the GWAS Catalog matched. Direction of effect was determined based on each SNP's odds ratio or beta value, and depending on each SNP's effect allele, the direction of effect was flipped. P-value was determined by a binomial test.

rates across East Asia and closely shared ancestry with other East Asian populations, we relied on genetic data from the other East Asian groups to investigate our hypothesis (18). Surprisingly, East Asia had lower risk allele frequencies on average for SNPs significant in gastric cancer risk from the GWAS Catalog (**Figure 3**). In comparison with EAS's mean allele frequency of approximately 0.394, EUR had the highest mean allele frequencies, with a mean difference of -0.056 (mean allele frequency of 0.45), followed by AMR with a mean difference of -0.055 (mean allele frequency of 0.449), AFR with a mean difference of -0.043 (mean allele frequency of 0.437), and SAS with a mean difference of -0.050 (mean allele frequency of 0.444). As for SNPs with higher risk allele frequencies in EAS, rs6039695, rs1057941, and rs11560253, nearest to the genes *ANKEF1*, *MTX1*, and *SNX13*, respectively, had the largest positive allele frequency difference with the other populations. We conducted a paired t-test, which showed that the mean differences between EAS and populations from other regions were statistically significant (EAS-EUR p=4.71x10^{-3}, EAS-AFR p=3.84x10^{-2}, EAS-SAS p=1.41x10^{-3}, EAS-AMR p=3.97x10^{-4}) (**Figure 3**). In addition, we conducted a comparison using the tag SNPs identified from the remapping analysis, which showed that while East Asians still had a lower mean risk allele frequency compared to other populations, the extent of these differences was attenuated and became non-significant in general (EAS-EUR p=0.116, EAS-AFR p=0.124, EAS-SAS p=4.1x10^{-2}, EAS-AMR p=9.3x10^{-2}) (**Figure 4**).

DISCUSSION

We sought to investigate the genetic risk factors behind high gastric cancer incidence in populations of Korean ancestry or East Asian ancestry generally and what effects SNPs identified in non-Korean populations have in Koreans. We predicted that East Asian populations would have higher gastric cancer risk allele frequencies, and that SNPs associated with gastric cancer in studies on non-Korean populations would have similar effects in Koreans. By conducting three analyses on gastric cancer GWAS data from the KoGES GWAS and the GWAS Catalog, we observed similarities and dissimilarities with regard to genetic architecture between a Korean-ancestry population and populations of non-Korean ancestries. A comparison of direction of effect between Koreans and other population ancestries suggested some transferability, and we were able to identify numerous SNPs that have stronger associations to gastric cancer in Korean populations based on previously identified associations. Additionally, the role of genetics in the high gastric cancer risk in East Asia appears limited or in need of further investigation to identify causal variants.

Our analysis of the GWAS SNPs' direction of effects determined that the majority of SNPs (68.42%) had a concordant direction of effect between the GWAS Catalog and KoGES (19). Since more SNPs show the same direction of effect, the significant SNPs reported in the GWAS Catalog appear to be transferable to an extent in the Korean GWAS. Therefore, this observation supports the idea of some shared genetic risk factors between previously studied populations and Korean-ancestry populations for gastric cancer.

From the remapping analysis, 120 SNPs were identified as more significantly associated with gastric cancer in the Korean-ancestry cohort than in previously identified SNPs in the GWAS Catalog (20). This remapping analysis can also be described as a regional association scan, which differs from formal fine-mapping by identifying more significant tag variants for a given phenotype rather than determining a confident set of candidate causal variants. We observed that no original (GWAS Catalog SNPs in the KoGES GWAS) or newly identified SNPs were statistically significant for association with gastric cancer risk based on the GWAS significance threshold, possibly due to limited sample size of the KoGES GWAS. The GWAS Catalog SNPs themselves are likely tag variants that have significant associations with gastric cancer due to proximity to causal variants through LD (21). Thus, the SNPs we identified are probably tag variants that are more closely correlated with causal variants for gastric cancer in the Korean population than previously identified SNPs.

The two allele frequency comparison analyses revealed an unexpected finding: East Asians had the lowest risk allele frequencies for SNPs significantly associated with gastric cancer risk in the GWAS Catalog and newly identified SNPs from the remapping analysis among the five 1000 Genomes continental populations. Our expectation was that because East Asia has higher gastric cancer incidence, a genetic contribution would mean higher allele frequencies for these variants. However, this was not the case. The two allele frequency comparisons differ somewhat, with the remapped SNPs being most likely closer to the allele frequencies of causal variants in Koreans. Despite these differences, both results argue against the hypothesis that genetic factors contribute meaningfully to the higher gastric cancer incidence in East Asian-ancestry populations. This conclusion, however, is limited by not considering the effect sizes with the frequency difference (i.e., polygenic score). There are other possible explanations for our negative result: the causal variants for gastric cancer may have different allele frequencies than tag variants in the GWAS Catalog, and the GWAS Catalog may not have the relevant SNPs responsible for the difference in genetic risk in East Asians. We observed there were several

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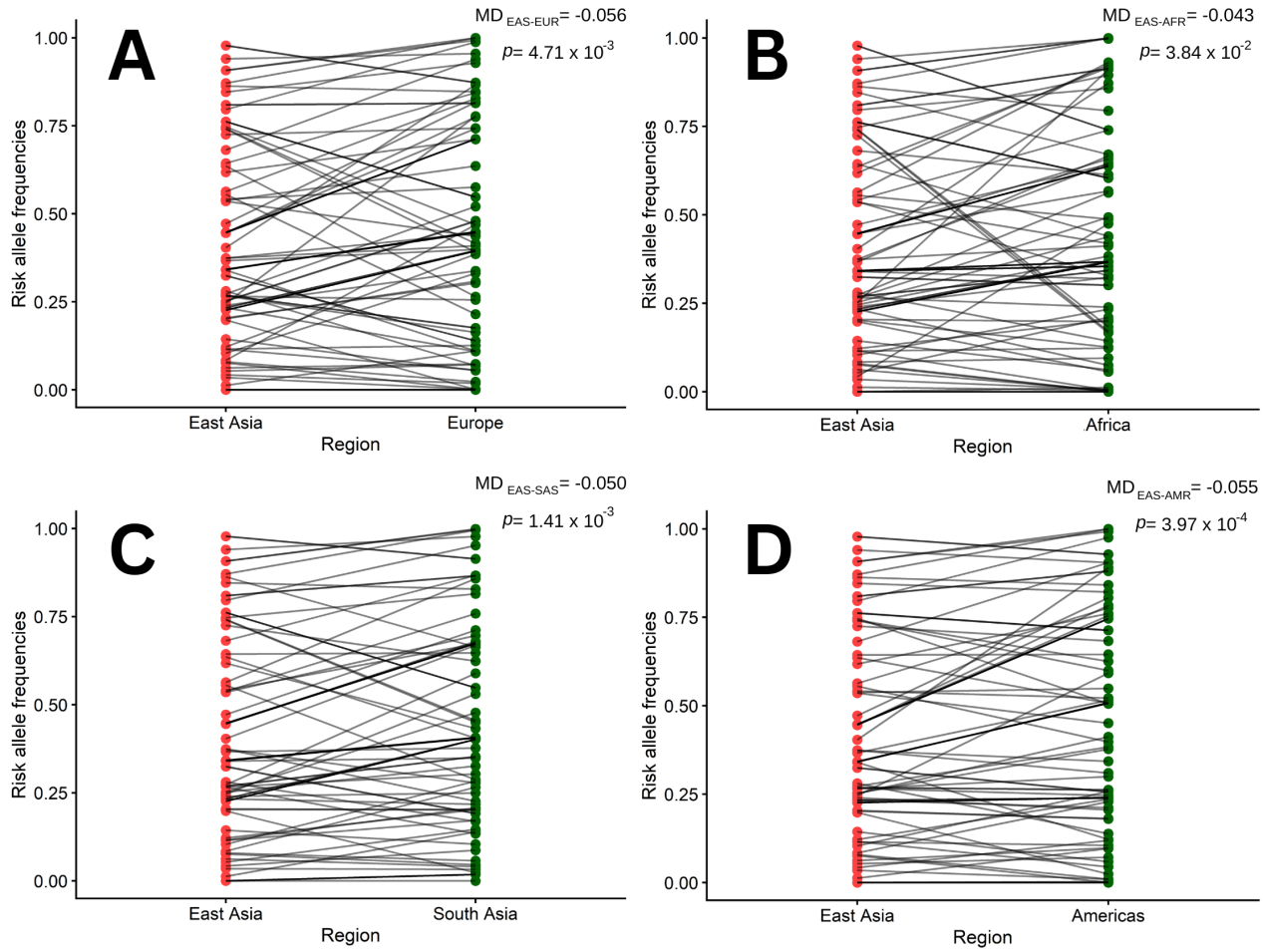

Figure 3: Mean risk allele frequency comparisons with East Asia for Europe, Africa, South Asia, and the Americas. Risk allele frequencies of GWAS Catalog SNPs in East Asia (red) compared to the corresponding risk allele frequency in other regions (green) for each SNP. East Asia is compared to Europe (A), Africa (B), South Asia (C), and the Americas (D). Mean difference (MD) from East Asia of risk allele frequencies for populations from Europe, Africa, South Asia, and the Americas is shown.

SNPs with very low allele frequencies in the Korean cohort, which might weaken a true signal because they are most likely not relevant to genetic difference in risk alleles.

Alternatively, environmental factors could be driving the high gastric cancer incidence rates observed in East Asian populations. *H. pylori* is a well-known cancer-causing pathogen (22). South Korea had a *H. pylori* seropositivity of 51% in 2015 compared to the crude global prevalence of 43.9% from 2015 to 2022 (23, 24). Additionally, high salt diets have been found to increase the risk of gastric cancer and may encourage carcinogenic effects of *H. pylori* (25, 26). South Korea had the 15th highest salt consumption levels in 2023, with 12.3 grams of daily salt consumption per capita (27). Heavy alcohol consumption is an additional factor that could contribute to a gender difference in gastric cancer incidence in Korea, possibly working in conjunction with the genetic missense variant rs671 at *ALDH2* to increase gastric cancer risk (10, 28, 29).

Limitations in this study include the lack of use of effect sizes, the incorporation of which might better reflect the differences in genetic risk across populations. For example, even though the average risk allele is not higher in frequency in East Asians, some variants could have outsized contributions to genetic risk if they have moderate to large impacts on gastric cancer and are higher frequency in East Asians. For the comparison of risk allele frequencies between populations, we did not consider effect sizes to calculate polygenic scores because the data from the GWAS Catalog comes from a collection of studies. This situation makes it methodologically challenging to combine the effect sizes estimated from different studies given potentially different methodologies and phenotyping across cohorts. Nevertheless, it would be ideal to calculate polygenic scores to jointly account for frequency and effect size differences. In addition, while the 1000 Genomes Project does not directly sample from a Korean-ancestry population, genetic variation in Koreans is mostly shared with other East Asian populations (18).

In summary, our analyses explored whether known genetic risk factors identified in previously studied populations were shared in Korean-ancestry populations and could explain disparities in gastric cancer. Our results suggest a limited role for genetics in the high gastric cancer incidence in Korean populations, but they also emphasize the need for larger studies of gastric cancer in East Asian populations, which would enable more accurate genetic predictions for gastric cancer for individuals of Korean and East Asian ancestry broadly.

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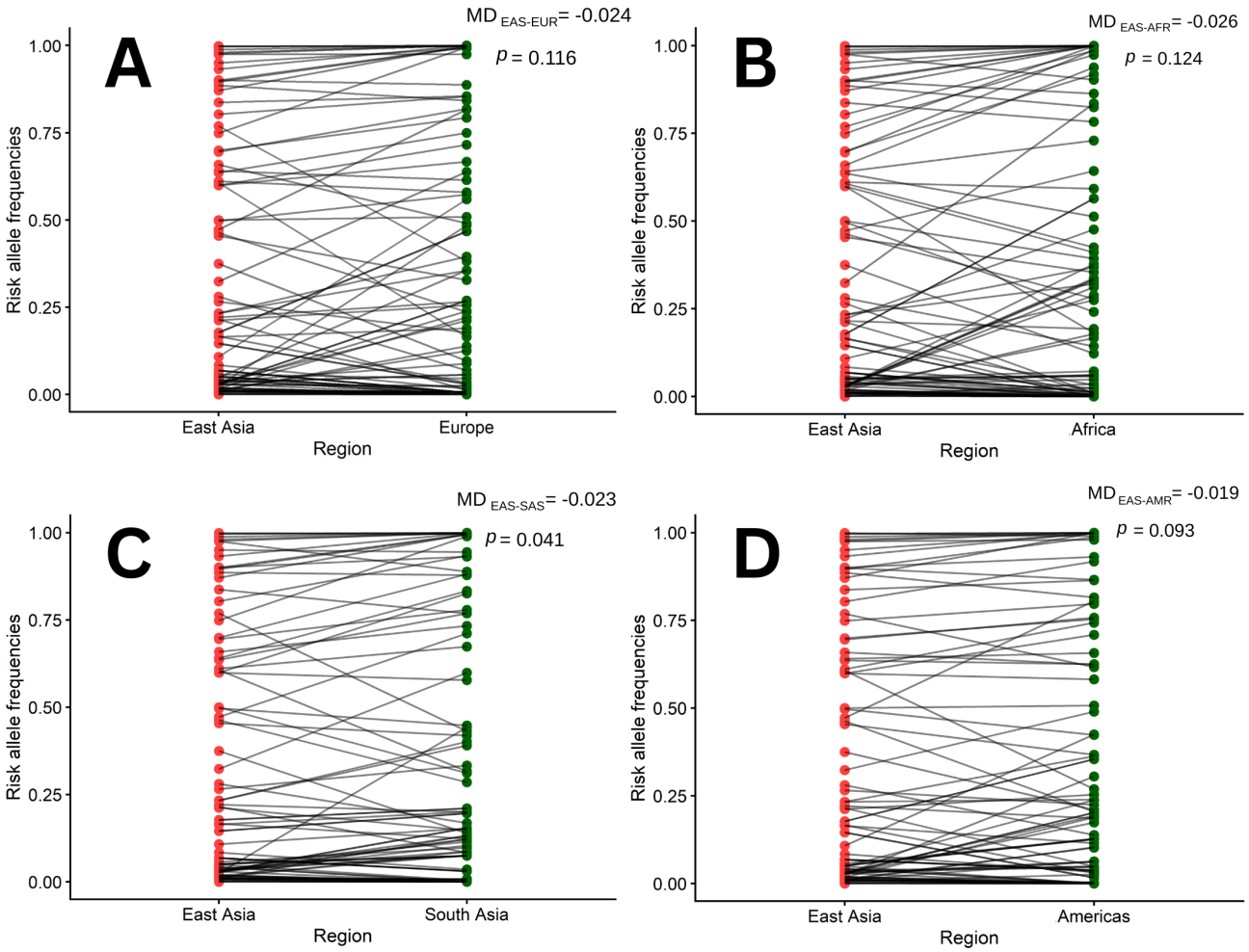

Figure 4: Mean risk allele frequency comparisons using newly identified SNPs with East Asia for Europe, Africa, South Asia, and the Americas. Risk allele frequencies of newly identified SNPs from the remapping analysis in the KoGES in East Asia (red) compared to the corresponding risk allele frequency in other regions (green) for each SNP. East Asia is compared to Europe (A), Africa (B), South Asia (C), and the Americas (D). MD from East Asia of risk allele frequencies for populations from Europe, Africa, South Asia, and the Americas.

MATERIALS AND METHODS

Dataset overview

We gathered data from two publicly available datasets. First, the GWAS Catalog, a regularly updated database of significant SNP-trait associations from published studies, was accessed on January 15th, 2025 and utilized to identify a list of previously identified SNPs significantly associated with gastric cancer (30). SNPs identified in studies of Korean populations (51 out of 173) were removed from the dataset (32, 33, 34). Second, a GWAS conducted on 72,298 individuals from the KoGES Consortium yielded summary statistics for 7,982,452 SNPs, which include statistical significance and estimated effect sizes (34). We used the data from KoGES_GCA, in which 378 individuals had gastric cancer and the other 71,920 individuals were controls. Using the programming language R, we conducted the three analyses described below with the code available in the appendix.

Remapping analysis

First, we conducted a remapping by retrieving a list of 122 SNPs observed to have a significant association with gastric cancer from the GWAS Catalog. We then located these SNPs in the KoGES GWAS through their rsID identifiers, ending up with a list of 106 SNPs. Each of these SNPs from the GWAS Catalog will have a different observed *p*-value in the KoGES GWAS. We identified SNPs with lower *p*-values in the KoGES GWAS compared to the original *p*-values of each of the GWAS Catalog SNPs within a range of 50,000 base pairs. We then recorded the SNP with the lowest *p*-value among the newly identified SNPs for each original SNP.

Direction of effect analysis

Second, the direction of effect analysis utilized the beta values or odds ratios (OR) of the gastric cancer-associated SNPs, which is a value included in the data from the GWAS Catalog. Based on these parameter estimates, we determined the direction of effect, whether the SNP increased or decreased gastric cancer risk, of each SNP. Beta values less than 0 decreased gastric cancer risk, while beta values more than 0 increased gastric cancer risk. OR values less than 1 decreased gastric cancer risk, while OR values more than 1 increased gastric cancer risk. If the effect allele in the KoGES GWAS differed from the effect allele in the GWAS Catalog, the observed direction of effect for that specific SNP in the

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KoGES GWAS was flipped. The direction of effect observed in the GWAS Catalog and the direction of effect observed in the KoGES GWAS were then compared for each SNP, yielding a percentage of SNPs where the direction of effect matched or didn't match between the GWAS. To assess the statistical significance of the results, we conducted a binomial test with a significance threshold of 0.05.

Allele frequency comparison analysis with GWAS Catalog SNPs

Third, we compared risk allele frequencies for five continental populations for gastric cancer-associated SNPs from the GWAS Catalog. The comparison utilized data from the 1000 Genomes Project to gather risk allele frequencies in Africa, the Americas, East Asia, Europe, and South Asia (17). Populations included in the East Asia data set were from China, Japan, and Vietnam. Since the alternate allele frequencies were provided in the VCF file from 1000 Genomes, we flipped them accordingly depending on whether the risk allele was the alternate or reference allele to obtain the risk allele frequency. The frequencies were then averaged for each region and compared to the average risk allele frequency for East Asia. We then conducted a paired t-test to determine if the differences between the risk allele frequencies for East Asia and the other regions were statistically significant based on a significance threshold of 0.05. We used the same procedure for the allele frequency comparison of the new tag SNPs identified in the remapping analysis.

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APPENDIX

https://github.com/lennayoon1018-cpu/Genetics-of-Gastric-Cancer-in-South-Korea