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Pharmacogenomic characterization of 14,490 Korea individuals based on the Korea Biobank Array v2.0

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Pharmacogenomics, a key component of precision medicine, seeks to improve drug safety and efficacy by accounting for inter-individual genetic differences. Although pharmacogenomics is globally recognized as a key component of precision medicine, research efforts have largely focused on European populations. This imbalance has limited the relevance of current guidelines for East Asians, including Koreans, who possess distinct allele frequencies and haplotype structures. This is largely because population-specific studies and tools for discovering pharmacogenomic variants in East Asians remain limited. To address this gap, we analyzed pharmacogenetic variants in 14,490 Korean individuals from the Korean Genome and Epidemiology Study (KoGES) using the Korea Biobank Array v2.0 (KBAv2.0). Phasing and imputation were performed with a reference panel constructed from whole-genome sequencing (WGS) data of 8,062 Korean individuals. We identified 759 pharmacogenomically loci, of which 96 (12.648%) were polymorphic sites. All individuals carried at least one pharmacogenetic variant, with an average of 10.163 variant loci per person (range: 3–21), underscoring the universal relevance of genotype-informed prescribing. As a result of comparison in predicted phenotypes with other populations, CYP2C19 intermediate metabolizer phenotype was more common in Koreans (47.142%) than in Western populations (28.806%), suggesting the need for ancestry-based clopidogrel dosing. In VKORC1, the Variant Present phenotype (rs9923231) was found in 99.358% of Koreans, but only 13.793% of Africans, indicating a substantial difference in warfarin dose requirements. These findings support the critical need for ancestry-informed pharmacogenomic guidelines to ensure optimal drug efficacy and safety for precision medicine implementation.