

# Large-scale study of East Asian individuals reveals a number of previously overlooked genetic variants

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Broad, population-based investigations known as genome-wide association studies (GWAS) are now a standard tool for helping scientists to pinpoint genetic variations that can contribute to disease risk or pathology. However, most of the studies performed to date have

focused predominantly on populations of European ancestry, and therefore ignore or overlook risk markers that specifically predominate among other ethnic groups. A recent GWAS from a large team of scientists based in Korea and Japan, including Yukinori Okada of the RIKEN Center for Genomic Medicine in Yokohama, has addressed this inequity by specifically seeking out factors that might contribute to metabolic disease in East Asians (Fig. 1).

In a GWAS, scientists analyze [genomic data](#) from large numbers of people who manifest a particular condition or trait of interest. They do this by seeking out small sequence changes known as single-nucleotide polymorphisms, or SNPs, that show a strong statistical association with the presence or absence of that particular trait.

In an earlier study, Okada and colleagues analyzed more than 14,000 Japanese individuals and found several previously unidentified loci of interest. This time, they performed an initial analysis in over 12,000 Korean individuals, and then replicated apparent ‘hits’ in a far larger group of over 30,000 individuals from Japan, Korea and China. This increased scale gave the researchers the ability to identify rare but meaningful associations with greater confidence.

They focused on finding genetic variants associated with imbalances in blood sugar, cholesterol and other indicators of metabolic function. “The recent rise of prevalence of metabolic diseases like diabetes, hyperlipidemia and chronic renal disease is a serious medical problem,” says Okada, “and these types of studies have been mainly conducted in European populations, but there are few studies on Asians.”

The team’s investigation uncovered 33 SNPs associated with metabolic function, 10 of which were previously unidentified. One of these [SNPs](#) was closely linked with variability in blood sugar levels, although the same SNP showed no significant association with this metabolic trait in a

northern European cohort. The researchers also identified a segment of chromosome 12 that appears to affect multiple metabolic phenotypes in both Europeans and East Asians; however, the specific sequence variations associated with these traits differ between the two populations.

Collectively, these data highlight the importance of expanding the breadth of GWAS analyses to cover the full spectrum of ethnic diversity. Okada and colleagues are planning to embark on an even larger-scale GWAS of East Asian populations in the near future.

**More information:** Kim, Y.J. et al. Large-scale genome-wide association studies in east Asians identify new genetic loci influencing metabolic traits. [Nature Genetics](#) 43, 990–995 (2011).

Kamatani, Y., et al. Genome-wide association study of hematological and biochemical traits in a Japanese population. [Nature Genetics](#) 42, 210–215 (2010).

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