Large genomic study finds tri-ancestral origins for Japanese population

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Fine-scale genetic structure of the modern Japanese and its three ancestry origins. (A) Geographic regions in Japan from which the samples were recruited are described. (B) PCA analysis based on common variants with a minor AF (C) Rare variant–based PCA-UMAP analysis (D) ADMIXTURE analysis with K set to 3. (E) UMAP1 is negatively correlated with the fraction of K2 ancestry. Credit: Science Advances (2024). DOI: 10.1126/sciadv.adi8419
A multi-institutional team of geneticists and genomic and genotyping specialists in Japan has sequenced the genomes of thousands of Japanese people from across the country, looking to settle the debate surrounding the ancestry of the Japanese people.

In their study, published in the journal *Science Advances*, the group sequenced the genes of so many people that they were able to decode the tri-ancestral origins of the Japanese population—and also to identify gene variants that may make some Japanese people more likely to develop certain illnesses.

The research team began by noting that most large-scale genetic sequencing research efforts have been conducted on people of European descent, which, they point out, leaves gaps in knowledge surrounding Asian ancestral roots.

So they embarked on one of the largest non-European sequencing research efforts to date. They sequenced the genomes of more than 3,200 people living in seven regions in Japan and Okinawa to create the new Japanese Encyclopedia of Whole-Genome/Exome Sequencing Library (JEWEL).

The researchers found that modern Japanese people are descended from only three main ancestral groups: Jomon hunter-gatherers of the Neolithic, predecessors of the Han Chinese, and a yet-to-be identified group of people from Northeast Asia. Their findings contradict theories that the Japanese people are descendants of the Jomon and later migrants from continental Asia.

The research team also found modern Japanese people have genes from both the Denisovans and Neanderthals, some of which have been associated with the development of certain diseases, such as diabetes type II, prostate cancer, coronary heart disease and rheumatoid arthritis.
They note that the identification of such variants could lead to better health care for Japanese people.

The team's efforts lay the groundwork for future genomic studies of people of Asian ancestry by providing a centralized database (JEWEL) that can grow and be used by researchers all across the region.


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