# Neanderthal DNA contributes to genetic diversity, bringing more understanding to human evolution

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Credit: C0 Public Domain

The advent of DNA sequencing has given scientists a clearer insight into the interconnectedness of evolution and the web-like path that different organisms take, splitting apart and coming back together. Tony Capra, associate professor of biological sciences, has come to new conclusions about the influence of Neanderthal DNA on some genetic traits of modern humans.

The article "Neanderthal introgression reintroduced functional ancestral alleles lost in Eurasian populations" was published in the journal *Nature Ecology & Evolution* on July 27.

The ancestors of all modern humans lived across the African continent, until approximately 100,000 years ago when a subset of humans decided to venture further afield. Neanderthals, an extinct relative of modern humans, had been longtime residents of Europe and central and south Asia; their ancestors had already migrated there 700,000 years previously. The humans who moved into central Asia and the Middle East encountered and reproduced with Neanderthals. Neanderthal DNA is present in some modern humans, and now research shows that can sometimes be a good thing.

"When Neanderthals split off from what became the human population 700,000 years ago, they took specific genetic variants along with them. Some of these genetic variants were later lost in human populations. We show that interbreeding with Neanderthals restored hundreds of thousands of previously lost genetic variants," said Capra. "These reintroduced genetic variants are more likely to have positive effects than genetic variants unique to Neanderthals."

In practice these reintroduced variants might have helped to regulate negative traits associated with Neanderthal DNA including autoimmune and neuropsychiatric diseases and addiction risk. Connecting how genetics alter risk is essential to understanding the function and development of disease.

"With this research we identify a unique set of very old genetic variants that predate Neanderthals, but that may have enabled segments of Neanderthal DNA to remain in the DNA of modern humans," said David Rinker, the first author of this research and postdoctoral scholar in the Capra Lab. "Pinpointing when those alleles (i.e., variant forms of genes) originated along the human timeline offers an evolutionary perspective on which genetic variants keep modern humans healthy, and has broad implications for how disease risk factors have evolved."

Capra's lab worked with data from the 1000 Genomes Project and the Neanderthal Genome

Project, two open initiatives that document genetic variation in detail. The researchers collaborated with Emily Hodges, assistant professor of biochemistry, to conduct a functional dissection of Neanderthal and human DNA to identify which variants have functional effects. "This analysis gives physical proof of our hypothesis," noted Capra. "It serves as a blueprint for doing analyses of this kind on a larger scale because we've proved the effect of these reintroduced genetic variants on a molecular level."

**More information:** David C. Rinker et al. Neanderthal introgression reintroduced functional ancestral alleles lost in Eurasian populations, *Nature Ecology & Evolution* (2020). DOI: 10.1038/s41559-020-1261-z

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