Lasting effects of Neandertal DNA on gene expression in modern humans

Michael Dannemann
Kay Prüfer
Janet Kelso

Video Abstract

Keywords: Neandertal introgression, Gene expression regulation, Protein sequence variation, Human evolution

Posted Date: February 25th, 2021

DOI: https://doi.org/10.21203/rs.3.rs-275885/v1

Abstract

Neandertals may be long gone, but their genetic legacy lives on in modern humans. If you are of non-African descent, approximately 2% of your genome is composed of Neandertal DNA – a consequence of interbreeding between these hominids and early humans more than 50,000 years ago. Despite the extensive amount of time passed, this event appears to have a lasting effect. Recent work has revealed that Neandertal DNA influences modern human traits such as skin pigmentation and strengthened immunity, but the precise consequences of this interbreeding are still being worked out. Now, scientists at the Max Planck Institute in Germany have found evidence that Neandertal DNA modifies the expression of certain genes and may even be associated with some genetic diseases. Scanning the genomes of hundreds of people living today, the researchers were able to precisely identify and locate Neandertal DNA fragments in modern humans. Using this genetic map, they looked for associations between these fragments and gene expression levels in 48 human tissues. So, are genes today still being affected by this ancient DNA? The study found that while the amount of Neandertal DNA in modern genomes has decreased over recent human history, the lingering ancient DNA appears to have a large effect on the regulation of nearby genes. The researchers show that the Neandertal DNA that persists in modern humans has contributed to both protein sequence differences and to regulating gene expression levels. In fact, hundreds of human genes were found exhibiting expression differences that are linked to these archaic genomic regions. Interestingly, the team found multiple instances of Neandertal DNA associated with the risk of developing disorders such as Crohn's and Parkinson's disease. This study shows that, though it occurred quite some time ago, the genetic exchange between humans and Neandertals has functional impacts on people today by modifying gene expression and protein sequences. The finding that remnants of this interbreeding may influence disease risk opens the door for further research into the genetic basis of these disorders.