Journal of Genetic Genealogy

Journal: www.jogg.info
Originally Published: Volume 12, Number 2 (Summer 2024)
Reference Number: 122.001

EDITOR'S CORNER

Author(s): J. David Vance

Journal of Genetic Genealogy

EDITOR'S CORNER

By J. David Vance

Another 2024 issue of the Journal hits the presses! While the steady stream of scientific articles is a welcome indicator of the continuing advances in genetic genealogy, we are also building up a library of case studies and examples of the application of genetic genealogy techniques on groups of varying sizes and family connections. One of the inspiring results of these examples for me is the differing approaches across autosomal, Y and mt DNA testing that authors have developed. If you or anyone you know are looking for help in applying genetic genealogy techniques, be sure to tell them to visit our Issue and Articles Archives and use the search and filter options to find real-life examples!

The cover of this Summer 2024 issue shows a "tube diagram" version of the draft pangenome reference for the human genome which was recently released by the Human Pangenome Reference Consortium (http://humanpangenome.org, a multi-institutional consortium working on the next official iteration of a reference for human chromosomes). This first draft reference was built from 47 individuals selected so the reference includes a representation of the diversity of the human genome as well as begin to document the larger structural variants that occur as currently-unmapped larger mutations in our chromosomes.

But building this diversity into the reference genome comes at a cost in complexity that we haven't accounted for yet in genetic genealogy – our reference genome is still the "official" (and largely static) hg38 reference. Unlike that reference where each chromosome can be represented by a single string of alleles, these new pangenomes have to be represented using more complicated structures like the "tube diagram" on our cover page.

So our cover page for this issue is both a warning and a promise that genetic genealogy will have to address this complexity in time. How will SNPs and STRs be represented under a pangenome reference? How will we handle mutations that occur on strands of DNA which themselves are only carried by a fraction of the population? And are structural variants stable enough to indicate shared ancestry and become as useful to us as other types of mutations?

We live indeed in interesting times…