

In This Issue of JoGG

In this second issue of the Journal of Genetic Genealogy (JoGG), completing Volume 1, we have three original articles, one of each of the types of articles that JoGG publishes, regular articles, review articles, and brief communications.

The article on the “Pitfalls of Determining Haplogroup F*,” by Whit Athey, describes an example of the type of small study that is now possible for “amateur geneticists” to carry out, given the availability of all of the new test offerings from commercial labs. In this article the point is made that when SNP results are not really compatible with STR results, one should not automatically believe the SNP results—at least until they have been confirmed in a second round of testing.

Ian Logan has written a review of the medical implications of whole-genome mtDNA sequencing. In contrast to the HVR1 and HVR2 results with which we’ve become familiar, one should not order a full mtDNA sequence without understanding that medically important information may come with the results. There is not only a privacy issue, but also there are undoubtedly many people who would not like to learn of their possible susceptibility to a disease. Others will likely be of the opinion that whatever problem may potentially exist, they would like to know about it. It will be very important to think about which camp you fall into before ordering the test. This article also shows several examples of mutations that are potentially problematic, and discusses in general terms how one may

interpret the medical implications of any particular mutation that shows up in your results.

Dr. Logan’s estimates that 5-10% of people who have the full mtDNA sequence may have a “medically significant mutation,” which is a surprisingly high percentage. However, it should be remembered that most of the medically significant mutations will not necessarily lead to actual expression of a disease, but only to increased susceptibility.

The brief communication for this month is another small study organized by amateurs, but with important implications for the Y phylogenetic tree. This article by Whit Athey and Ken Nordtvedt shows that the haplogroup, I-M223, does not form a new subgroup of I (called I2 in the Y chart from Family Tree DNA of November, 2004), but instead it should remain as a subgroup of I1, namely I1c, as first proposed by its discoverers.

Ann Turner continues her regular column for JoGG entitled “Satiableness Curiosity,” designed to connect the needs and resources of the genealogy community with those of the professional geneticists. Her current column addresses “The Case of the Ubiquitous 16519C.” In this column she examines the reasons for this mutation showing up in so many mtDNA haplogroups. Is it simply because the CRS sequence has a rare polymorphism at this site, causing “differences from CRS” to occur almost everywhere, or have many different independent mutations occurred at this site?