

Reply to van Oven: Suggestions and caveats for naming mtDNA haplogroup

We agree with van Oven (1) that a uniform mitochondrial (mt) DNA haplogroup nomenclature is indispensable for the future studies, and we actually propagated the same idea a few years ago (2). The current conflicts regarding the Asian haplogroup nomenclature (1) were essentially caused by publication delay: at the time when we submitted the manuscript (July 14, 2009), there was no conflict between the haplogroup system in our paper (3) and PhyloTree (mtDNA tree Build 5; <http://www.phylotree.org/>) to which we referred. To avoid similar problems in the future, a possible solution is that researchers in the mtDNA field indicate to van Oven (1) on submission of a paper that certain haplogroup names are being introduced, so that other researchers would at least not choose the same code for their newly identified haplogroups. The blocked names (possibly with some further information) could then be shown in the bulk mtDNA tree on that website.

The haplogroup nomenclature as a uniform naming system is also intended to reflect (sub)continental affiliation of the lineages by partly contiguous ranges of numbers and letters. For instance, one can easily memorize that haplogroups A–G and M7–M13 are prevalent in East Eurasians, whereas H–K are specific to West Eurasians and M2–M6 are specific to South Asians. Previous researchers have carefully named the newly identified M, N, or R subhaplogroups by using number slots for each broad region, which, however, turned out to be of insufficient size (especially in the case of South Asia, even after the creation of an additional slot from M30 to M40). Unfortunately, this naming strategy was not generally realized, and subsequently, it got blurred when an increasing number of basal lineages were named quite arbitrarily.

According to our unpublished data, there are plenty of unnamed basal lineages in Asia. To avoid similar conflicts regarding haplogroup nomenclature and help researchers give “proper” haplogroup names for the novel lineages, we propose that any future numbering of novel (basal) lineages should select numbers from geographically prescribed slots (Fig. 1). Even then, there may be exceptional cases where the subcontinental origin of a basal lineage

may seem ambiguous. Now that it turned out that what the lineage we called M16 (3) has a sister lineage south of the Himalayas (4) but apparently does not have related lineages in East Asia, adopting haplogroup M62 that encompasses both lineages (1) might be a better choice.

In cases of well-established haplogroups, renaming should be done under exceptional circumstances. A superhaplogroup should not be named based on a single moderately or highly variable site. So, in retrospect, the naming of M4 as based only on the hypervariable site 16311 was most unfortunate; deliberately broadening the scope of haplogroup M13 to an entity that would be supported merely by a single site, 6023, that is moderately variable (5) would lead to instability in the naming system. A potentially second variant, A15924G, cannot come for support here, because it admits a different equally parsimonious reconstruction.

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