



Did haplogroup M23 originate in Africa

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Abstract

Haplogroup M23 is an ancient M haplogroup found only in Madagascar and among African Americans. In this paper we discuss the genomic, archaeological, and historical evidence that indicates that M23 may have originated in Africa, and that haplogroup M probably expanded across Africa before the out of Africa exit by anatomically modern humans 60kya.

Introduction

The L3(M,N) mtDNA haplogroups are usually assumed to have been taken to Asia 60kya, when homo sapien sapiens migrated out of Africa (OOA)(1). After the OOA event some researchers believe that macrohaplogroup M diversified and was taken back to Africa as a result of a back migration(2).

This view concerning the demic diffusion of haplogroup M may need revision given the recent discovery of haplogroup 23 and the widespread distribution of the macrohaplogroup M across Africa(3). The discovery of haplogroup M23 suggest that we may have to change our views about the origin and spread of haplogroup M in Africa (4).

Results

All members of haplogroup M also have other well known differences

From CRS, namely 10398G and 10873C, which are the ancestral states, compared to the mutation 10398G>A and 10873C>T that occurred in Haplogroups N and R on the line to CRS, and the HVR1 mutation 16223T>C, which also occurred in haplogroup R, also on the line to CRS. Haplogroup M originated from an African Haplogroup L3 background.

L3(M) haplogroups are found throughout Africa. In addition to haplogroup M23 in Madagascar we also find other lineages belonging to the M macrohaplogroup including M*, M1, M3, M30, and M33 (see Map).

The M1 macrohaplogroup is found throughout Africa and Asia. The most widespread M haplogroup in Africa is M1. The basal M1 lineage has not been found outside Africa (6).

The Haplogroup M1 branch is defined by several mutations, including 195T>C, 16129G>A, 16249T>C and 16311 T>C, in the control region, and 6446G>A, 6680T>C, 12403A>C and 14110T>C (6). The RFLP of M1, considered diagnostic in many early studies, is by MnlI site loss at 12402 (an indicator of 12403T).

Gonzalez et al (7) reports that the highest frequency of M1 is found in Sub-Saharan Africa especially East Africa. The

molecular evidence makes it clear that haplogroup M1 is not confined solely to Ethiopia (4) as maintained by Olivieri et al (3).

In Tanzania the predominate M1 clades are M1, M1a1 and M1a5. In Senegal the predominate M1 lineage is M1c1.

In addition to haplogroups M1, M* and N in Sub-Saharan Africa we also find among the Senegambians hapotype AF24 (DQ112852. Gonder et al (7) maintains that LOd is "the most basal branch of the gene tree". The TMRCA for LOd is 106kya. This makes haplotype AF-24 much older than L3a.

Haplogroup M23 is a novel mtDNA lineage found among Malagasy(5). Haplogroup M23 is characterized by mutations 2706-8360, 9438, 9545, 10142, 10295, 11569, 11899, 12279, 12618 and 15025. The control region mutations of M23 are 152, 195, 204, 417, 533, 16263, 16311 and 16519. The M23 clade can be defined by coding regions np 10295 and control region transition at np 16223. The M23 control region mutations 152, 195, 16311, 16263 and 16519 are shared with haplogroup (hg) M1.

Discussion

Ricault et al (5) believes that hg M23 should be placed at the root of macrohaplogroup M. These researchers suggest that M23 is probably of South Asian origin.

This hypothesis may not be correct because no individuals with the M23 control region motif have been found in Asia. The only individuals sharing the diagnostic HVSI mutations of M23 are three African-Americans and one individual from the Arabian Peninsula (5). No individuals possessing M23 has been found in Africa up to now.

The presence of hg M23 among African-Americans is highly suggestive of an African, rather than Asia origin for hg M23. This results from the reality that hg M23 is predominately found among the Mikea group of Madagascar (5).

The aboriginal population of Madagascar was African. Mikea is the name for hunter-gatherer people who live in the forest(9). The Mikea, are descendants of Malagasy population that fled into the jungles to escape slave traders. During the Indian Ocean and Atlantic slave trades millions of Africans from East Africa and Madagascar were sold as slaves (9).

The ancestors of the African-Americans that share the haplogroup M23 coding variants probably came from region of East Africa and Madagascar. American slave traders collected slaves in east African and Madagascar and sold them throughout the United States (10). They took slaves from Madagascar and East Africa because the British Royal African Company monopolized the West African slave trade (10).

In 1680 the first Madagasy slaves and slaves from up the Mozambique Channel were sold in New England (10). According Mannix and Cowley (10) Governor Bradstreet of the State of Massachusetts in 1680 observed that the Malagasy

slaves were "betwixt forty or fifty Negroes, mostly women and children" (p.65). This is most interesting because it indicates that many female Malagasy female slaves were sold in America who could have deposited hg M23 in the United States.

The Royal African Company (RAC) was upset about the trade in Malagasy and Mozambique slaves. Mannix and Cowley (10), report that in 1683 the RAC claimed that as many as 900 slaves were transported to the United States every two months (p.243). In 1721, the RAC began to import slaves from Madagascar and Mozambique.

Using the figures of the RAC for 1683 (6x900) of 5400 slaves sold in the United States by American slave traders, we can conservatively estimate that between 1683-1721, for example, around 205,000 slaves entered the United States from Madagascar and Mozambique. This trade in Malagasy slaves did not end after the RAC began importing slaves from Southeast Africa, according to Mannix and Cowley(10) Malagasy slaves were still coming into the United States as late as 1806 (p.244).

The large number of African slaves from Mozambique and Madagascar explains the discovery of African Americans sharing the M23 haplogroups with the Malagasy. It is probably found among African-Americans and Arabians due to the large importation of Mozambican and Malagasy slaves during the Indian and Atlantic slave trades. It may be difficult to find hg M23 in Africa today, due to the large depopulation of many parts of Africa during the slave trade. W.E.B. DuBois claims that over 100 million Africans died during the slave trade.

Haplogroup M23 is believed to be an extremely old M haplogroup. The estimated founder age of hg M23 is 62-73 kyr (95% confidence interval, 44-94kyr) (5). This date is far older than the date for the OOA event from Africa to Asia (2). The antiquity of hg M23 supports the early expansion of the haplogroup M across Africa before the OOA exit.

Gonder et al (8) argues that the TMRCA of mtDNA L3(M,N) and their derivatives is around 94.3kya. It was not until 60kya that the TMRCA of non-African L3(M,N) exited Africa. This was 30,000 years after the rise of L3 and LOd and predicts a significant period of time for anatomically modern humans (amh) living in Africa to spread L3(M) haplogroups across the continent. This would explain the early age for hg M23. The existence of the basal L3a(M) motif and the LOd haplotype AF-24 among Senegalese supports this view.

Gonder et al (8) claimed that LOd is exclusive to the southern African Khoisan (SAK) population. The presence of the ancient AF-24 haplotype among the Senegalese, that is absent in other parts of Africa, suggest a long-term population in the Senegambia that preserved this rare haplotype—that originated early in the history of amh.

Moreover, the existence of the L3a-M motif in the Senegambia characterized by the DdeI site np 10394 and AluI site np 10397 in haplotype AF24 (DQ112852) make a 'back migration' of haplogroup M to Africa highly unlikely, since this haplotype is associated with LOd. The first amh to reach Senegal carrying haplogroup M probably belonged to the Sangoan culture which spread from East Africa to West Africa probably between 100-80kya.

Conclusion

The L3(M) haplogroups are found throughout Africa. These lineages include haplogroup M23 in Madagascar, and haplogroups M*, M1, M3, M30, and M33.

Debut et al (10) believes that hg M23 originated in India. But there is no evidence of Indians migrating to Madagascar. On the otherhand, the linguistic, archaeological, and anthropological evidence support a recent migration of Dravidian speaking people carrying haplogroup M from Africa to India (4,11). The Dravidian speakers and Africans also share many Y-chromosome haplogroups (12).

The M23 control region mutations 152, 195, 16311, 16263 and 16519 are shared with hg M1, which is found in the Senegambia region in association with Senegambian haplotype AF24 [13]. The reality that AF-24 is a haplotype of haplogroup LOd makes it clear that this haplotype is not only an ancient human genome, it is also evidence that AF-24 probably did not originate in Asia, since it was found among the Senegalese, and reflects an early migration from East Africa to West Africa. The presence of the nucleotides characteristic of macrohaplogroup M in Africa and the reality that M23 probably does not descend from an Asian M macrohaplogroup because of the absence of AF24 in Asia as noted by Sun et al (6), suggest that the expansion of M was probably from Africa to Eurasia. The existence of haplotype AF-24 and the LOd lineage in East and West Africa also implies the probable existence of the Proto-haplogroup M lineage in Africa, not Eurasia. One of these Proto-M haplogroups may be hg M23.

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