LETTERS

Chadic languages and Y haplogroups

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The January 2009 publication in this journal of an article entitled 'Human Y chromosome haplogroup R-V88: a paternal genetic record of early mid Holocene trans-Saharan connections and the spread of Chadic languages', by Cruciani *et al.*¹ represents a major step forward in our understanding of the African Y haplogroup diversity and pre-history. Approximately one year ago I published a detailed review of the earlier studies in this field.² Similar to Cruciani *et al.*, a major theme of my consideration was the probable link between the dispersal patterns of Y haplogroups and Afro-Asiatic languages. In particular, although the major focus of that review was upon Y haplogroup E1b1b1 (E-M35), similar to Cruciani *et al.*, I suggested that in the specific case of speakers of the Chadic family of languages, the high frequency of R haplotypes, which are otherwise uncommon in most of Africa, seems to be relevant for judging the strength of the competing theories regarding the origins of this language group within Afroasiatic. A comparison with this new publication can help put its findings into useful perspective.

In my review I proposed that the Y chromosomal evidence seemed to be most compatible with Blench's³ theory that Chadic is ancestrally most closely related to Beja and Cushitic, and arrived in its current position from the east and the direction of Sudan and the Red Sea. At that time, the data seemed to be much less compatible with Ehret's more novel position that it arrived from the north and is most closely related to Berber.⁴ A key consideration is that R haplotypes in various forms are generally Eurasian, being found as far away as Siberia, and their concentrated presence this deep in Africa has long been considered as evidence of movement of people from the direction of Levant, although the timing and route have been difficult to determine.^{5,6}

Blench's proposal that Chadic arrived with pastoralism involving goats, a way of life that spread from the Middle East, probably initially along the eastern side of the Nile, therefore matches the DNA evidence very closely. Furthermore, I observed, as do Cruciani *et al.*, that the discovery by Černý *et al.* of a mitochondrial link between Chadic speakers and the Horn of Africa (Mitochondrial haplogroup L3f) also supports Blench's position.⁷ In summary, I proposed that, in this case, a Y haplogroup had possibly been carried by a wave of innovation in life style (pastoralism), independently of language or overall genotype. As I remarked:

The recent autosomal DNA study of Tishkoff *et al.* (2009)⁸ confirms that on the whole, Chadic speakers are more closely related to their Nilo-Saharan neighbours than to any other Afroasiatic group. Looking at the genome beyond Y-DNA these peoples show far less Eurasian ancestry than the Beja, for example.

Nevertheless, Cruciani *et al.* argue that the discovery of V88 now demonstrates a strong Y chromosomal link between Chadic speakers and speakers of other Afro-Asiatic languages to the north of the Sahara. Their key evidence for this is their new data for Siwa in Western Egypt, a Berber-speaking area having approximately

27% R-V88 (93 people tested). These data are surprising and should be seen as one of the major discoveries of the article, and requiring an explanation such as the one given by the authors.

The article contains a striking contour map, which shows a coloured band of R-V88 passing from Siwa down to the Chadic speaking area, which would match Ehret's proposed migration route. However, analysis of the article shows that this coloured band simply joins two areas with high frequency, Siwa and the Chadic area. There are of course no data for populations along this band, which runs through the Sahara. One single population is therefore very important in their account. The potential importance of gaps in the data should be considered in several other directions also.

The study also contains no data for Sudan, which lies between the Chadic speakers in the west, and the Afro-Asiatic speakers in the horn of Africa and near the Red Sea. There are also no data for eastern Egypt. These areas are critical in determining whether Blench is likely to be correct, because they represent the path along which he believes pastoralism spread. Data for Sudan are limited but, as the authors note, the 2008 paper of Hassan *et al.* does seem to indicate a potential presence of R-V88 there.⁹

We know from this and other studies that E1b1b1 dominates the Berber speaking area, leaving the Siwa result as quite surprising and unique. This in turn raises the question of what other surprising and unique populations may exist in other oases and unsampled regions.

With the limitations of the data in mind, it could perhaps be argued that Cruciani *et al.*'s hypothesis is strengthened by the similarity between their age estimate for the R-V88 clade and the time depth at which we know the Sahara was green, making movement in the area between Siwa and Lake Chad more practical. However, unfortunately, such age estimates are one of the most controversial parts of any study of this kind. Different estimation techniques are likely to give a wide range of possible answers.

CONFLICT OF INTEREST

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- 2 Lancaster A: Y Haplogroups, archaeological cultures and language families: a review of the multidisciplinary comparisons using the case of E-M35. *J Genet Geneal* 2009; **5**: 35–65.
- 3 Blench R: The westward wandering of Cushitic pastoralists. Explorations in the pre-history of Central Africa; in Baroin C, Boutrais J (eds): *L'Homme et l'animal dans le bassin du lac Tchad*. Paris: IRD Edn, 1999, pp 39–80.
- 4 Ehret C: *The Civilizations of Africa: A History to 1800*. Oxford: James Currey, 2002.
- 5 Cruciani F, Santolamazza P, Shen P *et al.*: A back migration from Asia to sub-Saharan Africa is supported by high-resolution analysis of human Y-chromosome haplotypes. *Am J Hum Genet* 2002; **70**: 1197–1214.
- 6 Luis JR, Rowold DJ, Regueiro M *et al.*: The Levant versus the Horn of Africa: evidence for bidirectional corridors of human migrations. *Am J Hum Genet* 2004; **74**: 532–544.
- 7 Fernandes V, Costa MD, Hájek M, Mulligan CJ, Pereira L: Migration of Chadic speaking pastoralists within Africa based on population structure of Chad Basin and phylogeography of mitochondrial L3f haplogroup. *BMC Evol Biol* 2009; **9**: 63.
- 8 Tishkoff SA, Reed FA, Friedlaender FR *et al.*: The genetic structure and history of Africans and African Americans. *Science* 2009; **324**: 1035–1044.
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Reply to Lancaster

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In January 2010, we published in this journal a report¹ on the frequency distribution of the Y chromosome haplogroup R1b1a (R-V88) in Africa, where it can be found at frequencies as high as about 90%. This haplogroup (or its ancestor) most likely traces its origins back to Eurasia, but is presently found very rarely outside Africa.

In our original publication,¹ we described two important patterns in the genotyping data. The first observation was that the highest frequencies of the R1b1a haplogroup were found among Afro-Asiatic-speaking populations from the Central Sahel, with Chadic mostly contributing to this pattern. We have now extended our analysis to a further 258 unrelated male subjects from northern Cameroon (Table 1). As can be seen from Table 1, the extended data fully confirm the pattern originally observed.

The second observation was regarding a genetic contiguity between the Chadic-speaking peoples from the Central Sahel and several other Afro-Asiatic-speaking groups from North Africa, including

Ouarzazate Berbers from Morocco, Mozabite Berbers from Algeria, Siwa Berbers and several Semitic groups from Egypt, and, possibly, different groups from Algeria,² Tunisia³ and Egypt,^{3,4} with R1b1a frequencies ranging from 1 to 3% in Algeria to about 4% in Tunisia, to 26.9% in the Siwa. We interpreted these data by suggesting that they are more compatible with Ehret's hypothesis, which proposes that Chadic peoples arrived from the North through the Sahara (the 'trans-Saharan' hypothesis),⁵ rather than with Blench's theory, which states that Chadic-speaking pastoralists reached the Chad Basin through the Sahel from an eastern Sudanic Cushitic-Chadic motherland (the 'inter-Saharan' hypothesis).⁶

Considering the mitochondrial DNA, the populations from the Chad Basin also show some genetic peculiarities when compared with other populations living south of the Sahara. Mitochondrial DNA haplogroups L3f⁷ and L3e5,⁸ which are uncommon in the sub-Saharan area, were found to be relatively frequent in the Chad Basin region, with estimated coalescence ages similar to those we obtained for the Y chromosome R1b1a haplogroup. The lineage L3f3 can be traced back over the millennia to L3f,⁹ and this led the researchers who analyzed L3f3 from the Chad Basin⁷ to propose ancient links between this haplogroup and Chadic-speaking peoples coming from East or North East Africa. However, the presence in North Africa of the supposedly autochthonous Chad Basin haplogroup L3e5^{8,10} would seem to suggest another possible scenario, which is more compatible with the 'trans-Saharan' migration route.

Table 1 Frequencies (%) of the Y chromosome R1b1a and R1b1a4 haplogroups in Central Africa

Population	Country	Linguistic affiliation ^a	N	Haplogroup			References
				R1b1a	R1b1a* (xR1b1a4)	R1b1a4	
Songhai	Niger	NS/Songhai	10	0.0	0.0	0.0	Cruciani <i>et al</i> ¹
Fulbe from Niger	Niger	NC/Atlantic	7	14.3	14.3	0.0	Cruciani <i>et al</i> ¹
Tuareg	Niger	AA/Berber	22	4.5	4.5	0.0	Cruciani <i>et al</i> ¹
Ngambai	Chad	NS/Sudanic	11	9.1	9.1	0.0	Cruciani <i>et al</i> ¹
Hausa	Nigeria (North)	AA/Chadic	10	20.0	20.0	0.0	Cruciani <i>et al</i> ¹
Fulbe from Nigeria	Nigeria (North)	NC/Atlantic	32	0.0	0.0	0.0	Cruciani <i>et al</i> ¹
Yoruba	Nigeria (South)	NC/Defoid	21	4.8	4.8	0.0	Cruciani <i>et al</i> ¹
Ouldeme	Cameroon (North)	AA/Chadic	24	95.8	95.8	0.0	Cruciani <i>et al</i> ¹ , this study
Mada	Cameroon (North)	AA/Chadic	17	82.4	76.5	5.9	Cruciani <i>et al</i> ¹
Mafa	Cameroon (North)	AA/Chadic	9	88.9	33.3	55.6	Cruciani <i>et al</i> ¹ , this study
Guiziga	Cameroon (North)	AA/Chadic	9	77.8	22.2	55.6	Cruciani <i>et al</i> ¹
Daba	Cameroon (North)	AA/Chadic	29	51.7	34.5	17.2	Cruciani <i>et al</i> ¹ , this study
Guidar	Cameroon (North)	AA/Chadic	9	66.7	22.2	44.4	Cruciani <i>et al</i> ¹
Massa	Cameroon (North)	AA/Chadic	7	28.6	14.3	14.3	Cruciani <i>et al</i> ¹
Mandara	Cameroon (North)	AA/Chadic	82	42.7	17.1	25.6	This study
Podowko	Cameroon (North)	AA/Chadic	5	80.0	20.0	60.0	This study
Other Chadic	Cameroon (North)	AA/Chadic	8	50.0	12.5	37.5	Cruciani <i>et al</i> ¹ , this study
Shuwa Arabs	Cameroon (North)	AA/Semitic	5	40.0	40.0	0.0	Cruciani <i>et al</i> ¹
Kanuri	Cameroon (North)	NS/Saharan	7	14.3	14.3	0.0	Cruciani <i>et al</i> ¹
Fulbe from Cameroon	Cameroon (North)	NC/Atlantic	76	18.4	10.5	7.9	Cruciani <i>et al</i> ¹ , this study
Moundang	Cameroon (North)	NC/Adamawa	21	66.7	14.3	52.4	Cruciani <i>et al</i> ¹
Toupouri	Cameroon (North)	NC/Adamawa	31	71.0	12.9	58.1	This study
Fali	Cameroon (North)	NC/Adamawa	105	21.9	17.1	4.8	Cruciani <i>et al</i> ¹ , this study
Tali	Cameroon (North)	NC/Adamawa	22	9.1	4.5	4.5	Cruciani <i>et al</i> ¹
Mboum	Cameroon (North)	NC/Adamawa	11	0.0	0.0	0.0	Cruciani <i>et al</i> ¹ , this study
Bamileke	Cameroon (South)	NC/Bantu	52	0.0	0.0	0.0	Cruciani <i>et al</i> ^{1,4} , this study
Bakaka	Cameroon (South)	NC/Bantu	12	0.0	0.0	0.0	Cruciani <i>et al</i> ^{1,4}
Ewondo	Cameroon (South)	NC/Bantu	32	3.1	3.1	0.0	Cruciani <i>et al</i> ^{1,14} , this study
Biaka Pygmies	CAR	NC/Bantu	33	0.0	0.0	0.0	Cruciani <i>et al</i> ^{1,14}

^aAA, Afro-Asiatic; NC, Niger-Congo; NS, Nilo-Saharan.

In his Letter, Lancaster¹¹ revisits our original data and provides valuable comments on our paper. Following his own previous review,¹² he argues that our interpretation may have been affected by poor population coverage in relevant regions from East and North East Africa. We agree that data from those areas that are particularly important in order to discriminate between the two theories (eg, Eastern Egypt and Sudan) would be very important. As far as we are aware, there are no data for Eastern Egypt. The investigation by Hassan *et al*¹³ in Sudan fills in the map of North East Africa, at least in part, by providing Y-chromosome haplogroup data on additional relevant population samples. However, the power of these data is limited by the low level of resolution, as no R1b1 internal markers were analyzed. Furthermore, even if one assumes that all the R1b1 Y chromosomes found in Sudan harbor the V88 mutation, there are little data to support the hypothesis that these chromosomes are the product of an ancient migration from the East. The highest frequencies of R1b1 chromosomes from Sudan were observed in the Hausa, a Chadic-speaking population that has migrated from the West, and in the Copts, a population group that is known to be largely the result of recent migrations from Egypt over the past two centuries.¹³ By contrast, only two R1b1 chromosomes were found among the Beja in Sudan,¹³ confirming our previous results that Chadic-speaking populations are distinguished from Cushites, at least at the Y chromosome variation level.¹

In summary, currently available genetic evidence seems to favor our previous hypothesis¹ that the Y chromosome haplogroup R1b1a is a paternal genetic record of the proposed 'trans-Saharan' migration.⁵ It will be interesting to see how the proposed pattern develops as more detailed information about the phylogeographic structure of this haplogroup and a more refined method to estimate coalescence times become available.

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