McDONALD INSTITUTE MONOGRAPHS

Archaeogenetics: DNA and the population prehistory of Europe

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Published by:

McDonald Institute for Archaeological Research
University of Cambridge
Downing Street
Cambridge
CB2 3ER
(0)(1223) 339336

Distributed by Oxbow Books
United Kingdom: Oxbow Books, Park End Place, Oxford, OX1 1HN.
Tel: (0)(1865) 241249; Fax: (0)(1865) 794449
USA: The David Brown Book Company, P.O. Box 511, Oakville, CT 06779, USA.
Tel: 860-945-9329; FAX: 860-945-9468

ISBN: 1-902937-08-2
ISSN: 1363-1349

Edited for the Institute by Chris Scarre (Series Editor) and Dora A. Kemp (Production Editor).

Film produced by Gary Reynolds Typesetting, 13 Sturton Street, Cambridge, CB1 2QG.
Printed and bound by Short Run Press, Bittern Rd, Sowton Industrial Estate, Exeter, EX2 7LW.

Chapter 31

An Indian Ancestry: a Key for Understanding Human Diversity in Europe and Beyond

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A recent African origin of modern humans, although still disputed, is supported now by a majority of genetic studies. To address the question when and where very early diversification(s) of modern humans outside of Africa occurred, we concentrated on the investigation of maternal and paternal lineages of the extant populations of India, southern China, Caucasus, Anatolia and Europe. Through the analyses of about 1000 mtDNA genomes and 400 Y chromosomes from various locations in India we reached the following conclusions, relevant to the peopling of Europe in particular and of the Old World in general. First, we found that the node of the phylogenetic tree of mtDNA, ancestral to more than 90 per cent of the present-day typically European maternal lineages, is present in India at a relatively high frequency. Inferred coalescence time of this ancestral node is slightly above 50,000 BP. Second, we found that haplogroup U is the second most abundant mtDNA variety in India as it is in Europe. Summing up, we believe that there are now enough reasons not only to question a 'recent Indo-Aryan invasion' into India some 4000 BP, but alternatively to consider India as a part of the common gene pool ancestral to the diversity of human maternal lineages in Europe. Our results on Y-chromosomal diversity of various Indian populations support an early split between Indian and east of Indian paternal lineages, while on a surface, Indian (Sanskrit as well as Dravidic speakers) and European Y-chromosomal lineages are much closer than the corresponding mtDNA variants.

The title of this chapter was not chosen in order to insist that India was the place from where Europe was colonized by modern humans some 40,000-50,000 years ago. It does, however, imply that the understanding of population genetics of contemporary Indians is useful and needed in the attempt to reconstruct the process of the out-of-Africa spread of modern humans.

In our analysis below we largely restrict ourselves to the two sex-linked genetic systems: mtDNA and the Y chromosome. There is also extensive earlier literature on the genetics of Indian populations, based on the analysis of the distribution and frequencies of classical genetic markers (for reviews see Cavalli-Sforza et al. 1994; Papiha 1996). Yet the mere size of the Indian population, exceeding 1 billion (and much more if we include Pakistan and Bangladesh, i.e. the whole sub-continent) and its eth-

nic diversity make it clear that to comprehend the richness of the gene pool of Indians long-lasting systematic efforts are needed. Here we address a number of problems relevant to the placement of Indian genetic lineages within the context of the rest of Eurasia, taking the recent out-of-Africa colonization of the world by modern humans as a starting point.

The root

Most of the archaeological, anthropological and genetic evidence on modern humans (reviewed recently by Disotell 1999; Foley 1998; Harpending *et al.* 1998; Jorde *et al.* 1998) supports their recent origin in and spread out from Africa (Stringer & Andrews 1988). While this is generally accepted (but see also Wolpoff 1999), various alternative pathways and modes of this dispersal have been suggested (see, for example, Chu *et al.* 1998; Hammer *et al.* 1997; 1998; Jin *et al.* 1999; Kivisild *et al.* 1999a; Lahr & Foley 1994; 1998; Templeton 1997).

A route or routes

The peopling of Asia can be interpreted in terms of one or several pathways and also in terms of one, two or multiple migrations out of Africa. The third variable is time. The actual situation becomes even more complicated if there were migrations back to Africa, in between outward migration waves. First the alternative extremes: i) the northern route, over Sinai, leading to eastern Asia through the steppes of Central Asia and southern Siberia and ii) the southern route over southern Arabia, followed by the migration along the coastline of southern Asia (Fig. 31.1). While the northern route model could explain the peopling of the whole Eurasia by a single migration from Africa, the southern route model is interpreted as implying at least two separate Late Pleistocene dispersal events, one leading to the northwest and the other to the east of Eurasia (Cavalli-Sforza *et al.* 1994; Lahr & Foley 1994). On the first hypothesis, the molecular trees of present-day Eurasian populations are not expected to split at the depth of the time of their coalescence back to Africans. This means that the geographic sequestration can well be invisible in deeper branches and would start to appear in the terminal branches. This is because the eastern and western Eurasian populations should share the basic branches that were already present in the initial population that left Africa. Archaeologically, this model is expected to be expressed in largely uniform transitions in Palaeolithic tool technology. In contrast, the multiple dispersal models would predict the existence of more than one set of basic genetic lineage groups present in Eurasians. In archaeological terms this model does not foresee uniformity of technology associated with different waves of dispersal of modern humans (Foley & Lahr 1997).

Indians as a key

The geographic position of the Indian populations makes them a good example to study the question of the early dispersal of our ancestors. Unfortunately, it is not clear yet when the subcontinent became inhabited by modern humans. Archaeological and palaeoanthropological records are scanty and limited in details. The time period around 30,000–50,000 sr, when the first signs of modern humans can be traced in Eurasia (Smith *et al.* 1999) has revealed both Middle (up to around 20,000 BP) and Upper Palaeolithic (starting from c. 30,000 BP) tool assemblages in India (Joshi 1996). So far the earliest fragmentary skeletal evidence (at c. 34,000 C14 BP) of anatomically modern humans comes from Sri Lanka (Deraniyagala 1998). Note that this island was at that time connected with the continent.

In genetic distance trees based on classical genetic markers Indians cluster more closely with western Eurasian populations than with either other Asians or Africans (Cavalli-Sforza *et al.* 1994). This clustering supports the traditional classification of Caucasoids, according to which most Indian populations are included within this terminologicallysomewhat unsatisfactory, but widely used grouping, covering linguistically Indo-European-, Finno-Ugric-, Caucasian- and Hamito-Semitic-(Afro-Asiatic)speaking populations plus a few outliers, notably the Basques.

Two major waves of migrations into India have been proposed to account for this greater similarity of Indians with western Eurasians than with Mongoloid people to the east of India. The more widely known scenario is an invasion of nomadic Indo-Aryan tribes around 4000 BP either from the west or from the Central Asian steppes in the north. The other, more recently proposed hypothesis is based on the fact that some 8000–9000 years ago several varieties of wheat and other cereals reached India, presumably from the Fertile Crescent. This hypothesis is also supported by linguistically based suggestions of a recent common root for Elamite and Dravidic languages (Diamond 1997; Renfrew 1989).

We stress that these two hypotheses, which are not mutually exclusive, leave completely open the history of the 'indigenous', pre-Neolithic inhabit-

ants of India, and the question of their contribution to the gene pool of the contemporary Indians. Were they largely replaced by much more recent immigrants or alternatively, was the result of the recent migrations insignificant genetically while perhaps still profound culturally? There are currently about 500 tribal populations scattered over the Indian peninsula and generally thought of as the survivals of the pre-Neolithic Indians (Cavalli-Sforza *et al.* 1994; Papiha 1996). These tribal populations make up only a minor fraction of the total population of the present-day Indians. Study of their genetic identity would allow one to ask an important question: Do the tribal populations possess genetic lineages absent or rare among the other Indian populations or are they largely genetically identical to the latter, particularly if the differences attributable to drift can be clearly distinguished?

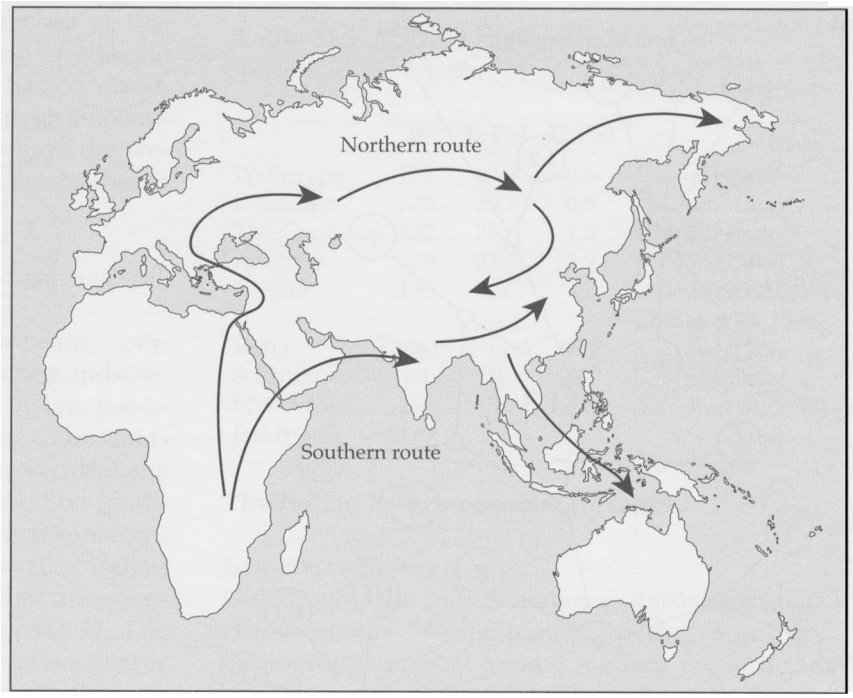

Figure 31.1. *Two altcrnative routesfor out-of-Africa migration.*

Our work: questions, populations and methods

The hypotheses set out above are the basis of our research in the form of two questions. The first is in the placement of the genetic lineages of Indians in the global context. We are trying to understand, in both qualitative and quantitative terms, the extent of the overlap of the genetic lineages found in India with those found elsewhere. Second, provided that such lineage clusters can be reliably reconstructed, can we establish when they diverge from those found elsewhere? Clearly the two questions are connected and the answers we look for depend on a detailed general knowledge about the genetic structure of the other Eurasians as well, and, at least partially, of the Africans.

So far we have analyzed 19 different Indian populations, covering the subcontinent geographically from Punjab and Kashmir to Sri Lanka and western Bengal. For a wider context, we have also investigated southern Chinese, Anatolian, Trans-Caucasian and eastern European populations. Our project has also included different Ethiopian populations.

Two principal experimental approches have been used: i) the first hypervariable segment of the control region of mtDNA was sequenced and 15 diagnostic RFLP markers were typed from the coding region for haplogroup affiliation; ii) a selection of bi-allelic Y-chromosomal markers were studied together with STR loci. Phylogenetic analysis was performed following the method of reduced median networks (Bandelt *et al.* 1995) using maximum parsimony as the guiding principle. The general schemes of mtDNA and Y-chromosome haplogroups are presented in Figure 31.2.

Mitochondria1 DNA

Previous studies have revealed that mtDNA lineage clusters (haplogroups)are specific to large geographic areas (Chen *et al.* 1995; Torroni *et al.* 1996; Wallace 1995). For example, nine mtDNA haplogroups (H, I, J, K, T, U, V, W and X) comprise about 95 per cent of the western Eurasian mtDNA pool, including Mediterranean Africa, whereas haplogroups M, B, F and A are specific for Mongoloid populations. The sub-Saharan African mtDNAs belong largely to a mtDNA supercluster L, further divided as L1, L2 and L3 (Watson *et al.* 1997). *L3* is at the root of nearly all mtDNA diversity found outside sub-Saharan Africa.

Haplogroup M as a clusfer of the proto-Asian maternal lineages

The most frequent mtDNA cluster found among Indian populations is haplogroup M (Bamshad *et al.*

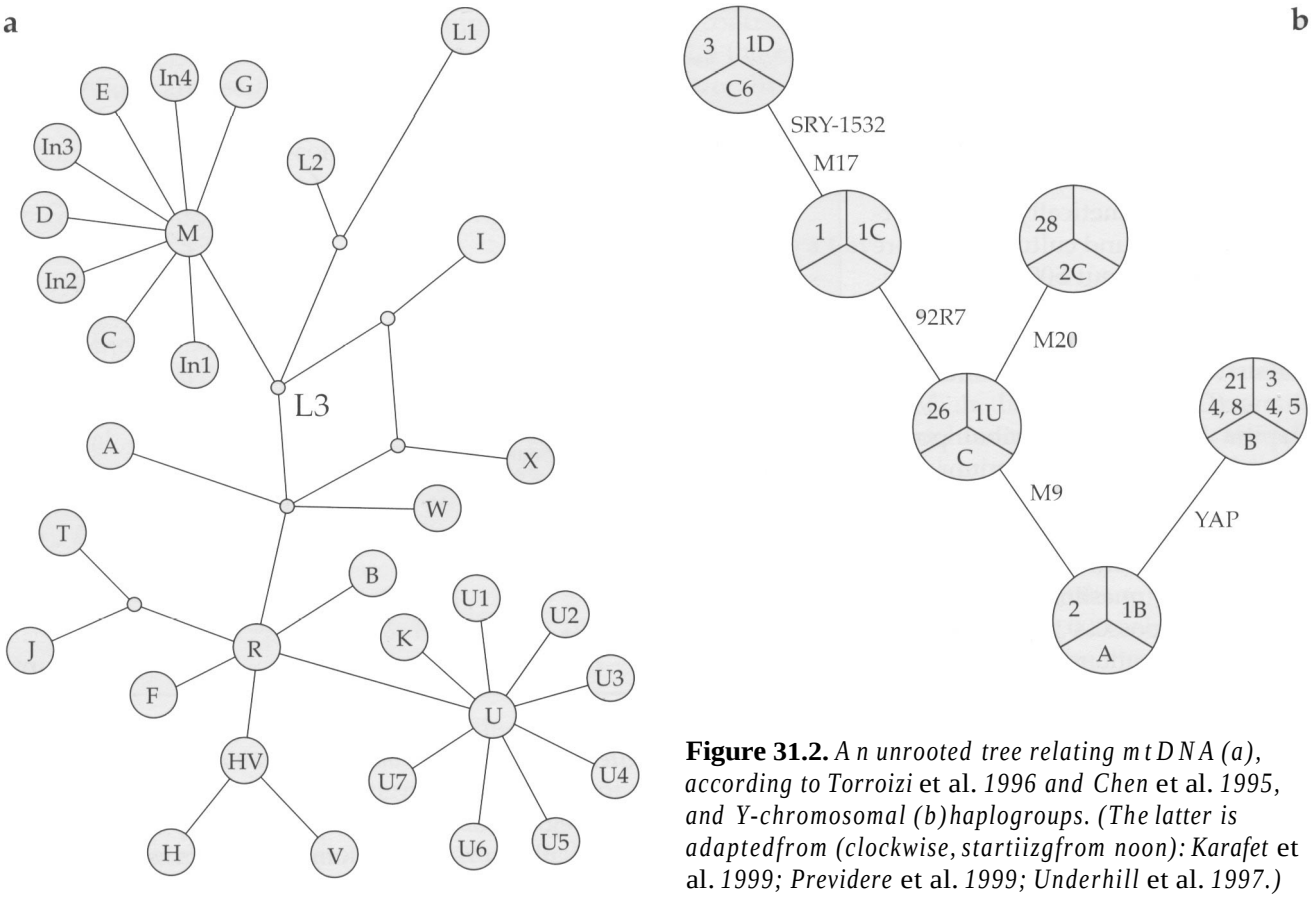

Figure 31.2. *An unrooted tree relating mtDNA (a), according to Torroizi* et al. *1996 and Chen* et al. *1995, and Y-chromosomal (b) haplogroups. (The latter is adaptedfrom (clockwise, startiizgfrom noon): Karafet* et al. *1999; Previdere* et al. *1999; Underhill* et al. *1997.)*

1997; Kivisild *et al.* 1999b; Passarino *et al.* 1996). This Asian-specific lineage cluster is widespread among Mongoloid populations (Kolman *et al.* 1996; Torroni *et al.* 1993b; 1994) as well as among Native Americans (e.g. Bonatto & Salzano 1997; Torroni *et al.* 1992; 1993a). Its frequency drops in southeastern Asians, where haplogroups B and F become dominant varieties of mtDNA (Ballinger *et al.* 1992; Wallace 1995). We have observed that the frequency of haplogroup M among Indians is even higher than among eastern Asians (Table 31.1), ranging from 40 per cent in Punjabis to almost a fixation level among Chenchus, a tribal hunter-gatherer community in Andhra Pradesh (97 per cent; $n = 96$).

There is an important finding, having possibly profound implications for the interpretation of the peopling of southern and eastern Asia, and allowing a better understanding of the genetic background of the populations living in Central Asia. This is that the Indian haplogroup M sub-structure (sub-clusters Inl–In4 in Fig. 31.2) differs profoundly from that observed in Mongoloid populations (Kivisild *et al.* 1999b). Two conclusions are immediately apparent: i) the expansion of the super-haplogroup M in Indians and Mongoloids, including the genesis of the region-specific sub-clusters, was clearly separated in space; ii) since then, there has been only very limited gene flow between India and eastern Asia. When did this separation happen? The haplogroup M coalescence time for eastern Asians has been estimated as around 55,000–73,000 BP (Chen *et al.* 1995). Coalescence calculations of Indian haplogroup M diversity (Kivisild *et al.* 1999b; Passarino *et al.* 1996) agree well with this estimate and suggest that the two macropopulations started to expand simultaneously, perhaps due to improved climate between the two major glaciations at about 73,000–65,000 and 24,000–16,000 years ago. The lack of any signs of significant re-migrations of eastern Asians to India is further supported by the scarcity of mtDNA lineages belonging to haplogroups A, B and F in India.

The fact that the Indian and eastern Asian/ Mongoloid varieties of haplogroup M are so distinct also allows one to interpret the haplogroup M lineages found in Central Asian populations phylogeographically: virtually all of those described so far

belong to the Mongoloid-specific branches of this haplogroup. This again tells us that no large-scale migration from Central Asia to India has occurred, at least any involving the presently Turkish-speaking populations of this area, among whom the frequency of haplogroup M is otherwise close to that in India and in eastern Asians.

Haplogroup L1 and the proto-western-Eurasian maternal lineages

The absence of haplogroup M in Europeans, compared to its equally high frequency among Indians, eastern Asians and in some Central Asian populations is inconsistent with the 'general Caucasoidness' of Indians. Any relationship between Indians and 'Caucasoids' must therefore be based on qualitative and quantitative data on genetic markers common to Europeans and Indians. Analyzing Indian maternal lineages further, we found that an extensive overlap was provided by haplogroup U. The distribution of haplogroup U (Table 31.1) is a mirror image of that for haplogroup M: the former has not been described so far among eastern Asians but is frequent in European populations as well as among Indians (Kivisild *et al.* 1999a). This reverse analogy goes further: Indian U lineages differ substantially from those observed in Europe and their coalescence to a common ancestor, like that for the haplogroup M lineages, dates back to about 50,000 years (Kivisild *et al.* 1999b). We infer from the fact that Indians and other populations do not generally share mtDNA lineages at the tips of the branches of the global phylogenetic tree with either eastern or western Eurasians that the Indian maternal gene pool has come largely through an autochtonous history since the Late Pleistocene.

The extent of a recent admixture in the Indian mtDNA pool

Our results suggest that the sum of any recent (the last 15,000 years) western mtDNA gene flow to India comprises, in average, less than 10 per cent of the contemporary Indian mtDNA lineages (Kivisild *et al.* 1999a). This fraction clusters closely together with the tips of the western Eurasian mtDNA tree, is higher among western Indian populations (Punjabis) and drops lower in most of the tribal populations studied by us. However, even the high castes share more than 80 per cent of their maternal lineages with the lower castes and tribals. We conclude that the recent enrichment of the Indian mtDNA pool with the western-Eurasian lineages is clearly detectable but had a relatively minor impact.

Table 31.1. *MtDNA haplogroup U and M frequencies worldwide.*

	n	U (%)	M (%)	
W-Europe	172	23.9	0.6	(our data)
E-Europe	329	23.7	0.6	(our data)
The Caucasus	532	23.6	1.5	(our data)
Anatolia	379	24.5	3.9	(our data)
C-Asia	195	8.7	41	(deduced from Comas *et al.* 1998)
India	1061	13	60.1	(our data)
S-China	69	0	37.7	(our data)
NW-Africa	268	13.7	0.6	(Rando *et al* 1998)
E-Africa	199	2.5	8	(our data)

The Indian Y-chromosomal lineages

East-west affinities

Unlike with the Indian maternal lineages, Indian Y chromosomes belong mainly to the same broad haplogroups present among modern western Eurasians (our unpublished data). In contrast, haplogroup 26 that is known to be highly frequent in eastern and southeastern Asians (Karafet *et al.* 1999), is very rare in Indians. At the same time, the Indian Y-chromosomal pool is rich in derivatives of haplogroup 26, such as 1 and 3 (1C and 1D in Karafet *et al.* 1999, respectively), which are, in turn, rare in the Chinese population.

However, an apparent lack of YAP+ chromosomes in Indians (Karafet *et al.* 1999; Bhattacharyya *et al.* 1999; Santos *et al.* 1999; Thangaraj *et al.* 1999; Underhill *et al.* 1997; our unpublished results) complicates the interpretation of largely western connection of the Indian Y chromosomes. It is well documented that in the European populations haplogroup 21 chromosomes harbouring the Alu insert are widespread, albeit generally at rather low frequency. The occurrence of the insert is more frequent in localities closer to India: the populations of the Middle East and the Turks carry this marker at frequencies about of 10 per cent or higher (Hammer 1994; Skorecki *et al.* 1997; Thomas *et al.* 1998). Yet Indians lack the YAP+ varieties, with the only exceptions being groups documented as being recent immigrants, the Siddis (Thangaraj *et al.* 1999) and Parsees (our unpublished observation). Several demographic scenarios may account for this discrepancy, including the possibility that the common pool of Y chromosomes shared by the western Eurasian and Indian populations lacked YAP+ chromosomes at the time of the split of the two proto-macropopulations.

In interpreting the Y-chromosomal data, we

must first recognize that certain haplogroups, frequent both among Europeans and Indians, are in fact complex aggregates of lineages deriving from central internal nodes of the Y-chromosomal phylogenetic tree. In the extreme case, haplogroup 2 is just an assemblage of lineages that so far cannot be assigned to any terminal node of the tree. Secondly, male and female genetic lineages might have been differently affected by gene flow, genetic drift or bottlenecks in their complex histories.

Returning to the beginning: almost certainly several routes

The main empirical conclusions from our results on Indians using M-haplogroup data are as follows:

1. MtDNA haplogroup M is, as a first approximation, equifrequent in populations living in three wide geographic areas: in southern, central and eastern Asia (+ Native Americans).
2. At the same time, the central and eastern Asian haplogroup M lineages belong to one, and the southern Asian lineages to another subset of haplogroup M.
3. The node — M* — is present both in southern and eastern Asia and the observed diversity implies a late Pleistocene origin.
4. Haplogroup M is virtually absent in western Eurasia and the lineages found so far in Africa belong all to a narrow sub-cluster, phylogenetically far apart from the ancestral nodal sequence.

Thus, the empirical results obtained so far clearly separate southern and eastern Asian populations from the western Eurasians through the sharing of haplogroup M. Its distribution combined with the age of its origin and diversification is best explained by a separate Late Pleistocene migration wave, most probably via the 'southern route'.

We note that the spread of haplogroup M to Eurasia by the southern route was during the final preparation of the manuscript suggested by an additional evidence relating African and Indian mtDNAs (Quintana-Murci *et* al. 1999).

The second set of conclusions is based on the phylogeography of the mtDNA haplogroup U:

1. Haplogroup U is absent in eastern Asian populations.
2. Haplogroup U is the second most frequent variety of mtDNA in India and in western Eurasia.
3. Indian and western Eurasian haplogroup U varieties differ profoundly; the split has occurred about as early as the split between the Indian and eastern Asian haplogroup M varieties. The data show that both M and U exhibited an expansion phase some 50,000 years ago, which should have happened after the corresponding splits.
4. Haplogroup U frequency is low in Africa with one dominant African-specific variety — U6.

These observations make it unlikely that the immediate ancestors of the carriers of haplogroups U and M left Africa as a common wave of migrants. We suggest that the ancestors of haplogroup U carriers used the 'northern route' because of U is present in both Indian and western Eurasian populations. Yet both these migrations seem to have occurred at comparable time depths.

The third set of conclusions build a bridge over Eurasia:

1. Topologically speaking, all major Caucasoid-specific mtDNA haplogroups (H, T, J and U) as well as the two major eastern Asian-specific haplogroups B and F, derive from a common internal node R*.
2. It seems that the only regions in Eurasia (and, indeed, globally) where one finds a large variety of the 'non-canonical' derivatives of this node are India and southern China. However, no data exist about the populations between these regions.
3. Nowhere is there any extensive overlap of western and eastern Eurasian mtDNA lineages except in Central Asian populations and Indians.

Our tentative general conclusion from the third list of conclusions is that the carriers of the R* node reached India very early and migrated eastwards, possibly together with the carriers of M*. As for M*, the expansion phase came later, leading in southeastern Asia to the formation of two major lineage clusters, F and B. In India, a large variety of derivatives of R* arose, while in western Eurasia its major derivatives that survived consist of haplogroups H, T, J and U. Among the latter, U seems to be the most ancient, explaining why one of its deepest branches is widely present in and specific for Indians.

For Y-chromosomal markers, we are limited to three empirical conclusions:

1. There is almost no direct overlap between the Indian and southeastern Asian (southern Chinese in our experiments) Y chromosomes.
2. The Y-chromosomal haplogroups present in Indians are also frequent in western Eurasians.
3. Not all Y-chromosomal haplogroups typical for western Eurasians are frequent in Indians.

Thus, we have observed a significant difference between the spread of Y-chromosomal and mtDNA markers in southern and southeastern Asians but we are not yet in a position to suggest whether these

differences are due to differential migrational patterns or, rather, due to profound differences in the demographic histories of the sex-specific genes, where the survival of the male lineages have undergone severe bottlenecks in some lineages and rapid bursts in others.

Returning now to the expansion of modern humans out of Africa, we suggest a scheme as shown in Figure 31.3. We regard it as minimalist, because many aspects known already are omitted, such like the sharing of mtDNA haplogroup U7 between Indian and Anatolian populations, lack of YAP+ Y chromosomes in India etc. However, data are lacking from such critically important regions as Afghanistan, Iran, Iraq, Myanmar, Thai etc. The ever-improving selection of genetic markers, combined with improvements in the understanding of the driving forces of demographic behaviour of humans and their prehistory and history in general, allow us to expect further progress in our reconstructions of the past. However, despite its shortcomings, we hope that this article shows that study of the Indian, mtDNA and Y-chromosomal lineages sheds new light on the general problem of the peopling of the Old World by modern humans.

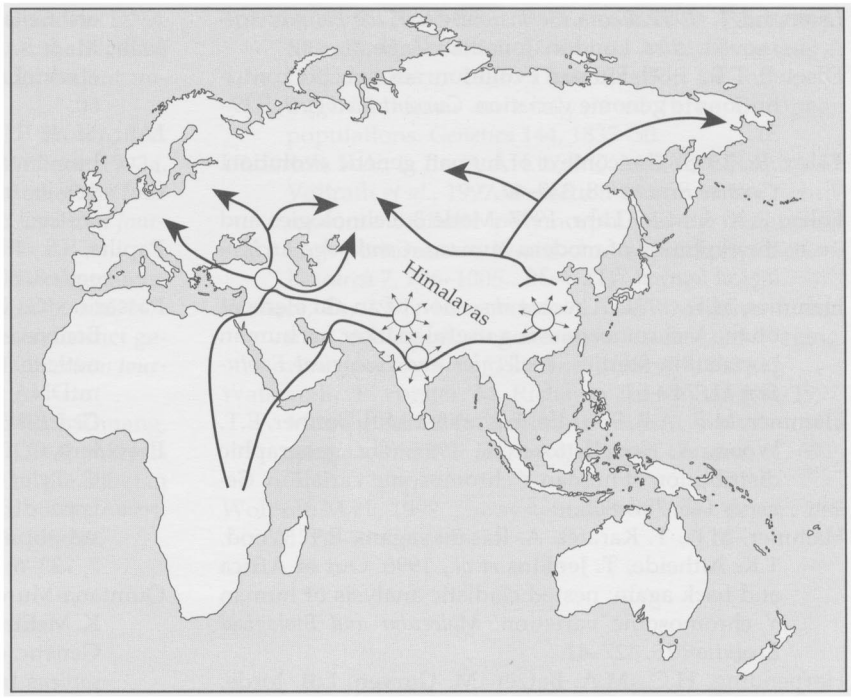

Figure 31.3. *Scheme combining the routes and expansions of modern human early dispersals.*

Acknowledgements

We thank Professor P. Broda and Dr P. Forster for many helpful suggestions in the preparation of this manuscript.

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