Chapter 35

Complex Signals for Population Expansions in Europe and Beyond

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François Jacob, in his brilliant 'The possible and the actual' (Jacob 1982), reminds us that 'scientific investigation begins by inventing a possible world, or a small piece of a possible world'. One may add that l Tz/F0 57O1Tfe piec ad

al. 1987; Vigilant *et al.* 1991), the last decade has demonstrated an increasingly better understanding of the phylogeny and phylogeography of mtDNA and of the Y chromosome. Here, the first influential achievement was a series of papers from Emory (reviewed in Wallace 1995) where, *inter alia*, it became obvious that human maternal lineages world-wide are very clearly structured geographically. This s coming from RFLP

analysis or the HVR sequence(s) alone were not informative enough to go further. Quite the opposite; it became clear that trees, based on HVR 1 sequence alone, were often phylogenetically wrong. However, a synthesis of what is known about polymorphisms in the coding region (extensive RFLP as a tool) and HVR (direct sequencing) removes most of the ambiguities and leads to a much better understanding of the details of the topology of the phylogenetic tree of mtDNA (e.g. Macaulay *et al.* 1999). This analysis owes much to the use of median networks as an approach (Bandelt *et al.* 1995).

In this contribution we demonstrateh provides new insi

graphic processes of the past and, in particular, allows to see informative differences there, where mere haplogroup frequency calculations are able only to register flat landscapes.

General

How much further can one go in resolution? It is obvious that 'the ultimate' answer lies in analyzing, in all collected samples, all 16,500 plus nucleotides of the mtDNA genome — to carry out total (high fidelity!) re-sequencing. There are now at least a thousand fully sequenced mtDNA genomes at hand and this body of data, although rather

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frequency exceeds that of

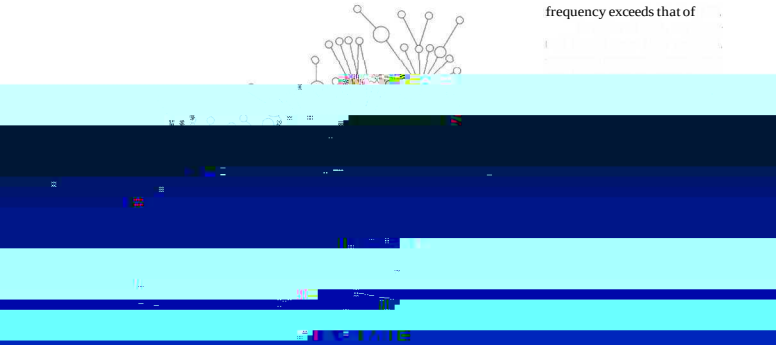
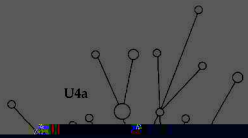


Figure 35.1. *Skeleton topology of the human mtDNA haphgroup U5 hypervariable 1 phylogenetic tree for western Eurasia. Circle sizes are proportional to numbers of individuals per haplotype. 'S' corresponds to a haplotype, most frequent among Saami population (HVR 1 motif 16,144; 16,189; 16,270).*

with a few interesting exceptions, more frequent in eastern Europe and is either absent or very rare in the Near East and elsewhere. In the European north, an interesting exception is the Saami mtDNA pool, where U4 is virtually absent.

We have constructed a HVRI-based phylogenetic tree for U4, using information from ~80 populations comprising a total of ~400 U4 genomes (Fig. 35.2). The topology of the U4 cluster is relatively simple, revealing the presence of a limited number of sub-founders. Of these, U4a and U4b are likely monophyletic, while U4c, determined by a transition at np 16,362, might be polyphyletic, at least in a pan-western Eurasian context. The highest frequencies of U4 (both in absolute terms and as a percentage of Hg U) can be observed actually not in Europe, but among Obi-Ugric Khantys and Mansis, living in northwestern Siberia. It is also frequent among the Finnic-speaking populations and in Volga Basin Turkic speakers, where, in some instances, its

U4a and U4b for the Germanic-speaking people (Germans, Norwegians, Swedes, Icelanders, Scots, German-speaking Swiss). Here the othe



recent debate on this issue (Simoni *et al.*, 2000; Torroni *et al.*,

with more recent events is that they might well be

the corresponding tree (at least for western Eurasia) is concerned, and that the problems identified here will be largely solved, or shown to remain ambiguous forever because of built-in limitations resulting from the length of mtDNA.

What is much less clear is how we can reach significantly better temporal resolutions. Take, for example, U5: a cluster coalescing around 40,000-50,000 BP but consisting, as we interpret it now, of a number of sub-founders coalescing about 12,000 BP. Even though U5 mtDNAs are frequent in the western Eurasian mtDNA pool, to identify numerous sub-clusters within it one does need to operate with large sample sizes. For less frequent mtDNA varieties, only very large data bases, consisting of data about tens of thousands of mtDNAs, will allow a detailed temporal analysis, in particular for a time

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