Chapter 8

MtDNA Markers for Celtic and Germanic Language Areas in the British Isles

In this context, the term 'marker' needs to be explained. Occasionally, in the course of the millennia, mutations may occur in the DNA of an organism, such as a human or a seed of grain, and these mutations are passed down to the descendants. If these descendants remain united by a common characteristic such as a language, a certain geographic area, or a resistance against cereal rust, the DNA mutation can be considered a marker for that characteristic, even though it does not cause the characteristic.

The two best-characterized genetic systems for identifying evolutionary markers in humans are the Y chromosome (passed down exclusively from the father to his male children) and mitochondrial DNA (transmitted exclusively from the mother to her children). Individuals living today differ genetically from each other as a consequence of different mutation events which occurred in their past ancestry, irrespective of whether these mutation events are interesting markers for any characteristic.

To date there have been two approaches to analyzing data for the exploration of genetic patterning: the summary method and the lineage approach. The summary method treats the entire data set in terms of a population concept which considers all the different genetic types as a single heritable un2is tvsingg)71Epsingg)71q0vsurredgg)71downredgg)71th (Torroni *et al.* 1993; Saillard *et al.* 2000). However, the European situation contrasts with that of America: modern languages and human DNA do not appear to correspond particularly closely. Geographic distance tends to be better at predicting how similar the DNA of any two European populations is (Rosser *et al.* 2000; Zerjal *et al.* 2001).

Nevertheless, it would be overly pessimistic to conclude that in Europe, genetic markers have no hope of shedding light on the prehistory of languages. Genetic markers tracing language migrations may well exist and these markers could then tell us about the routes and, via the molecular clock, the times of such migrations. However, unlike in North America, these markers may represent only a small minority of the gene pool, especially if the spread occurred by

et al. 2002). British data have been supplemented since by Capelli *et al.* (2003) who focus on potential Scandinavian contributions. In the

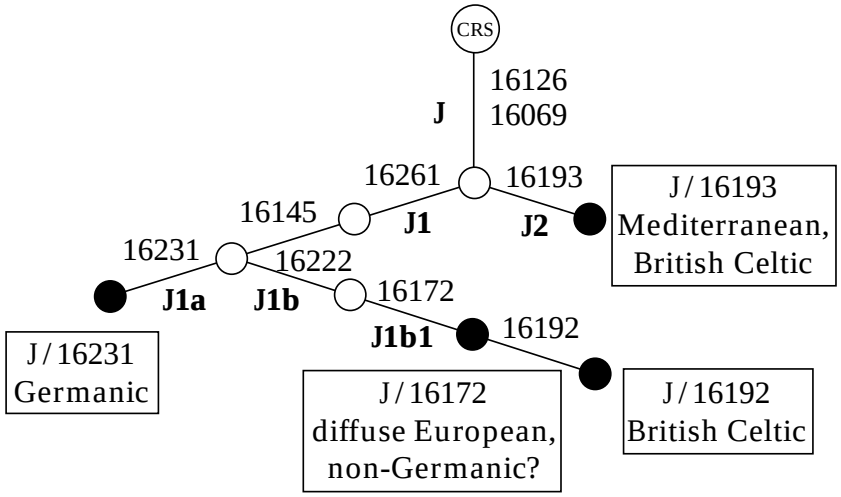

Figure 8.2. *Skeleton phylogeny of mtDNA type J. The numbers 16069, 16126 etc. refer to mutations at mtDNA nucleotide positions numbered as in Anderson* et al. *(1981). Labels J, J1 etc. refer to the branch nomenclature defined by Richards* et al. *(2000) and Richards & Macaulay (2000), whereas labels J/16193, J/16231 etc. refer to the provisional nomenclature used in this paper, as recommended by YCC (2002). Minor branches of J have been omitted here, and the geographic annotations are intentionally simplified. The mtDNA phylogeny and nomenclature is currently in flux due to complete mtDNA sequencing projects, so changes may be expected in the future.*

The probable reasons for this diffuse spread are twofold. Biologically, np16172 is prone to parallel mutations (L. Forster *et al.* 2002) and pseudo-matches may arise by new mutations. Statistically, the frequency of J / 16172 is generally very low on the Continent, making local frequency estimations even more erratic than in the other frequency maps presented here. Evidently, better sampling coverage of the

Figure 8.7. *Geographic distribution of mtDNA type J/16193. In the mtradius data base used for this search, 19,493*

worthy that Germans living close to the Dutch border harbour a low percentage of the Saxon marker (6/109 in the data of Pfeiffer *et al.* 1999) which is about as low as the English value of 3.5 per cent. Scant available mtDNA data from Jutland confirm a low frequency of the Saxon marker there, while the neighbouring Benelux countries and northern France, formerly home to the Frisians and Belgae, have not yet been studied for mtDNA.

Discussion

The traditional hypotheses on the arrival of Celtic-speakers and Germanic-speakers to the British Isles do not sit easily with the data from female-born mtDNA presented here. In the traditional, but not uncontested (Renfrew 1987) view, 'Celts' ultimately originating from a peri-Alpine Hallstatt/La Tène culture would have arrived in the British Isles around 600 BC. Neither this date nor an Alpine source area are entirely satisfactory from an mtDNA perspective. We tentatively place the arrival time for Insular Celtic mtDNA markers in the British Isles at thousands rather than hundreds of years BC. Furthermore the mtDNA profile of the 'Celtic' Alps is the opposite of the British Celtic profile as far as J is concerned: in the peri-Alpine region, Insular Celtic J types are absent or rare compared to the Germanic J/16231 marker which is clearly present in the Alps (see Fig. 8.8). As concerns the traditional view that northwest German Angles and Saxons arrived in Britain soon after the departure of the Romans around AD 410, our mtDNA survey reveals a paucity of the northwest German marker H/16189 in England. England does however yield an appreciable percentage of

descendants. Relative 'mutational' time is then converted to absolute time by multiplying it with the mtDNA mutation rate as estimated in Forster *et al.* (1996). Phylogenetic dating software (shareware) is available at www.fluxus-engineering.com. The mutation rate is the Achilles' heel for any absolute DNA chronology. Whereas relative genetic dates and their relative standard errors (both expressed in mutations) are by definition accurate, their conversion to accurate absolute dates (expressed in years) depends entirely on an accurate calibration of the mtDNA mutation rate. Should an improved mtDNA mutation rate become available in the future, all dates presented in this paper can be proportionately adjusted.

Acknowledgements

We thank Lucy Forster, Eleanore Conant and Marta Lahr (Cambridge) for providing access to unpublished mtDNA data, and Alda Ragalmuto and Valeria Chiavetta (Troina) for their technical assistance during data processing. We also thank Martin Richards (Leeds) and Mim Bower (Cambridge) for critically reading the manuscript.

References

Alves-Silva, J., M. da Silva Santos, P.E. Guimaraes, A.C. Ferreira, H.-J. Bandelt, S.D. Pena & V.F. Prado, 2000. The ancestry of Brazilian mtDNA lineages. *American Journal of Human Genetics* 67, 444–61.

Anderson, S., A.T. Bankier, B.G. Barrell, M.H.L. de Bruijn, A.R. Coulson, J. Drouin, I.C. Eperon, D.P. Nierlich, B.A. Roe, F. Sanger, P.H. Schreier, A.J.H. Smith, R. Staden & I.G. Young, 1981. Sequence and organisation of the human mitochondrial genome. *Nature* 290, 457–65.

Arnason, E., H. Sigurgislason & E. Benedikz, 2000. Genetic homogeneity of Icelanders: fact or fiction? *Nature Genetics* 25, 373–4.

Bandelt, H.-J., V. Macaulay & M. Richards, 2002. What molecules can't tell us about the spread of languages and the Neolithic, in *Examining the Farming/Language Dispersal Hypothesis*, eds. P. Bellwood & C. Renfrew. (McDonald Institute Monographs.) Cambridge: McDonald Institute for Archaeological Research, 89–97.

BBC, 2003. *Motherland: a Genetic Journey*. [Documentary on the ancestry of Jamaican descendants of African slaves.]

Bosch, E., F. Calafell, Z.H. Rosser, S. Nørby, N. Lynnerup, M.E. Hurles & M.A. Jobling, 2003. High level of male-biased Scandinavian admixture in Greenlandic Inuit shown by Y-chromosomal analysis. *Human Genetics* 112, 353–63.

Capelli, C., N. Redhead, J.K. Abernethy, F. Gratrix, J.F. Wilson, T. Moen, T. Hervig, M. Richards, M.P.H. Stumpf, P.A. Underhill, P. Bradshaw, A. Shaha, M.G. Thomas, N. Bradman & D.B. Goldstein, 2003. A Y-chromosome census of the British Isles. *Current Biology* 13, 979–84.

Dubut, V., L. Chollet, P. Murail, F. Cartault, E. Beraud-Colomb, M. Serre & N. Mogentale-Profizi, in press. MtDNA polymorphisms in five French groups: importance of regional sampling. *European Journal of Human Genetics*.

Faltings, V.F., A.G.H. Walker & O. Wilts (eds.), 1995. *Northwestern European Language Evolution*, Suppl vol. 12. Odense: University of Odense.

Forster, L., P. Forster, S. Lutz-Bonengel, H. Willkomm & B. Brinkmann, 2002. Natural radioactivity and human mitochondrial DNA mutations. *Proceedings of the National Academy of Sciences of the USA* 99, 13,950–54.

Forster, P., 1995. Einwanderungsgeschichte Norddeutschlands [Immigration history of North Germany], in Faltings *et al.* (eds.), 141–63. [English translation available at www.mcdonald.cam.ac.uk/genetics.]

Forster, P. & A. Toth, 2003. Toward a phylogenetic chronology of ancient Gaulish, Celtic, and Indo-European. *Proceedings of the National Academy of Sciences of the USA* 100, 9079–84.

Forster, P., R. Harding, A. Torroni & H.-J. Bandelt, 1996. Origin and evolution of native American mtDNA variation: a reappraisal. *American Journal of Human Genetics* 59, 935–45.

Forster, P., A. Torroni, C. Renfrew & A. Röhl, 2001. Phylogenetic star contraction applied to Asian and Papuan mtDNA evolution. *Molecular Biology and Evolution* 18, 1864–81.

Forster, P., F. Calì, A. Röhl, E. Metspalu, R. D'Anna, M. Mirisola, G. De Leo, A. Flugy, A. Salerno, G. Ayala, A. Kouvatsi, R. Villems & V. Romano, 2002. Continental and subcontinental distributions of mtDNA control region types. *International Journal of Legal Medicine* 116, 99–108.

Helgason, A., E. Hickey, S. Goodacre, V. Bosnes, K. Stefansson, R. Ward & B. Sykes, 2001. MtDNA and the islands of the North Atlantic: estimating the proportions of Norse and Gaelic ancestry. *American Journal of Human Genetics* 68, 723–37.

Hurles, M.E., C. Irven, J. Nicholson, P.G. Taylor, F.R. Santos, J. Loughlin, M.A. Jobling & B.C. Sykes, 1998. European Y-chromosomal lineages in Polynesians: a contrast to the population structure revealed by mtDNA. *American Journal of Human Genetics* 63, 1793–806.

Kivisild, T., S. Rootsi, M. Metspalu, S. Mastana, K. Kaldma, J. Parik, E. Metspalu, M. Adojaan, H.V. Tolk, V. Stepanov, M. Golge, E. Usanga, S.S. Papiha, C. Cinnioglu, R. King, L.L. Cavalli-Sforza, P.A. Underhill & R. Villems, 2003. The genetic heritage of the earliest settlers persists both in Indian tribal and caste populations. *American Journal of Human Genetics* 72, 313–32.

Macaulay, V., M. Richards, E. Hickey, E. Vega, F. Cruciani,

D.C. Wallace, 1996. Classification of European mtDNAs from an analysis of three European populations. *Genetics* 144, 1835–50.

Torroni, A., M. Richards, V. Macaulay, P. Forster, R. Villems, S. Nørby, M.L. Savontaus, K. Huoponen, R. Scozzari & H.-J. Bandelt, 2000. MtDNA haplogroups and frequency patterns in Europe. *American Journal of Human Genetics* 66, 1173–7.

Weale, M.E., D.A. Weiss, R.F. Jager, N. Bradman & M.G. Thomas, 2002. Y chromosome evidence for Anglo-Saxon mass migration. *Molecular Biology and Evolution* 19, 1008–21.

Wilson, J.F., D.A. Weiss, M. Richards, M.G. Thomas, N. Bradman & D.B. Goldstein, 2001. Genetic evidence for different male and female roles during cultural transitions in the British Isles. *Proceedings of the National Academy of Sciences of the USA* 98, 5078–83.

Y Chromosome Consortium, 2002. A nomenclature system for the tree of human Y-chromosomal binary haplogroups. *Genome Research* 12, 339–48.

Zerjal, T., L. Beckman, G. Beckman, A.V. Mikelsaar, A. Krumina, V. Kucinskas, M.E. Hurles & C. Tyler-Smith, 2001. Geographical, linguistic, and cultural influences on genetic diversity: Y-chromosomal distribution in northern European populations. *Molecular Biology and Evolution* 18, 1077–87.