

Report

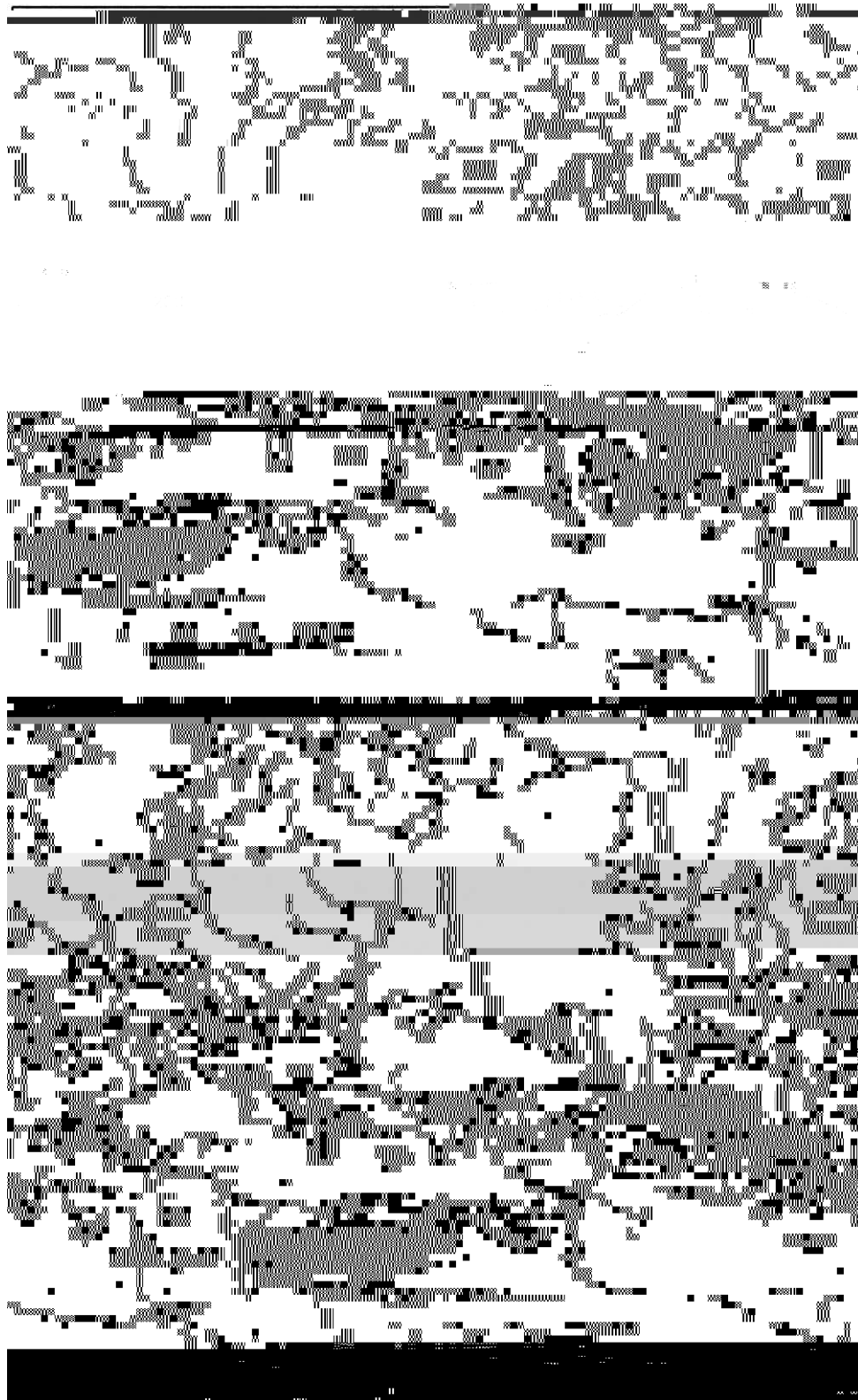


Figure 1 Regional map of Europe (modified from Gamble 1999, after Gamble 1986). *A*, Sample sizes for mtDNA data sets analyzed. *B*, Sample sizes for Y chromosome data sets analyzed. Note that, in the scheme we have used, Basques are the sole representatives of southwestern Europe; samples from France are grouped with northwestern Europe, and those from Galicia are grouped with the western Mediterranean.

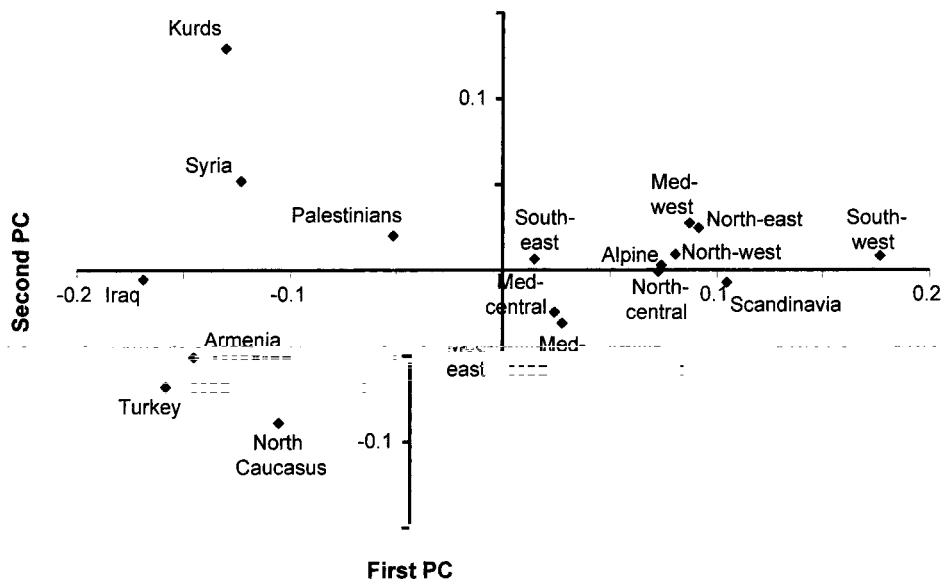


Figure 2

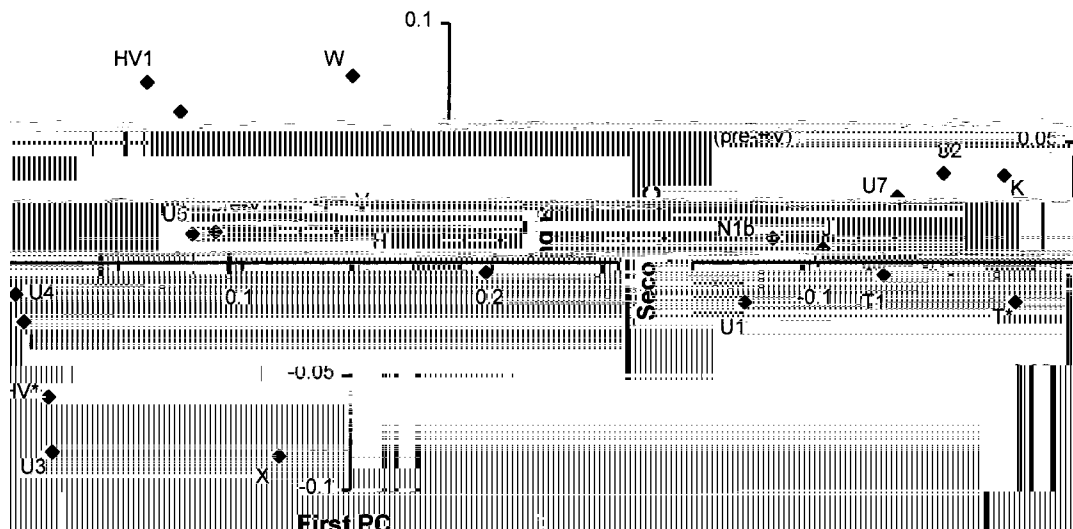


Figure 3 Plot of the contribution of each haplogroup to the first and second PC in the analysis of figure 2

nantly Near Eastern, with some (probably recent) gene flow along the Mediterranean, and haplogroups U5 and pre-V are predominantly European.

The southeast-northwest clines in classical marker frequencies have been interpreted, by comparison with radiocarbon evidence, as representing a substantial demic diffusion of Near Eastern farming communities into Europe in the early Neolithic period (Sokal et al. 1991; Cavalli-Sforza et al. 1994). However, the pattern in mtDNA haplogroup frequencies that we see here indicates similarity between Europeans and Near Easterners primarily in southeastern Europe and along the Mediterranean, whereas archaeological evidence would point to the main expansion of agriculture being into central Europe (Bogucki 2000; Price 2000). Thus, it seems rather unlikely that the pattern in mtDNA haplogroup frequencies could have been generated mainly by a Neolithic expansion.

A well-resolved Y chromosome gene tree now exists and can be found, along with all of the main haplogroup nomenclatures that have been proposed, in the recent article by the Y Chromosome Consortium (2002). Y

325(btng)-btinP230C-390(iapn)-343.8(90es)-375(90l).wJsece8itmoriculshowEu-

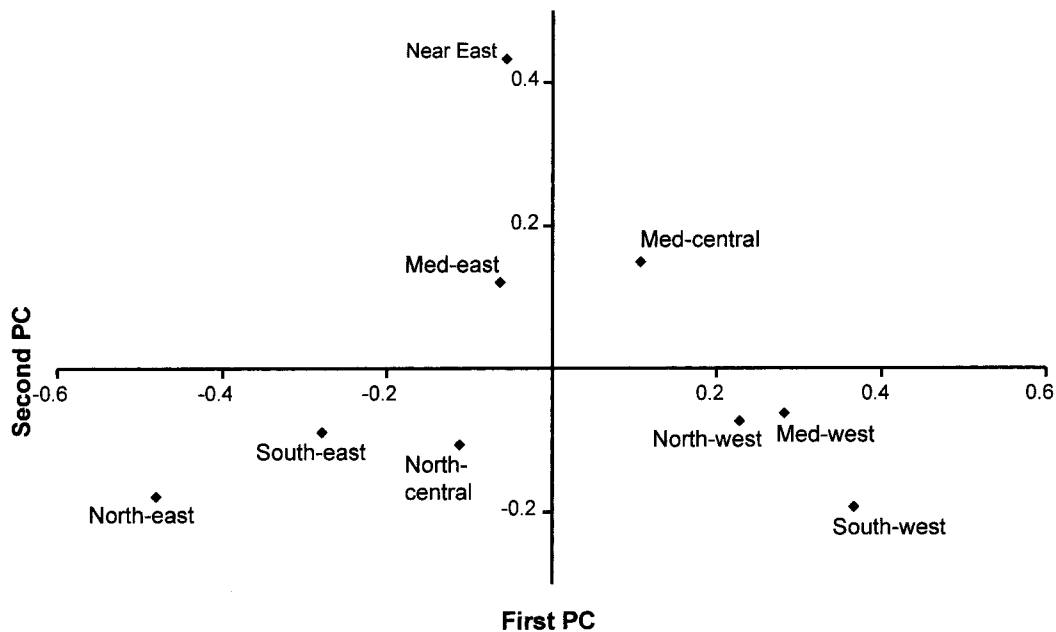


Figure 4 Region-based PC analysis of Y chromosome biallelic marker data (Semino et al. 2000), grouped into major haplogroups (Y Chromosome Consortium 2002). Med = Mediterranean.

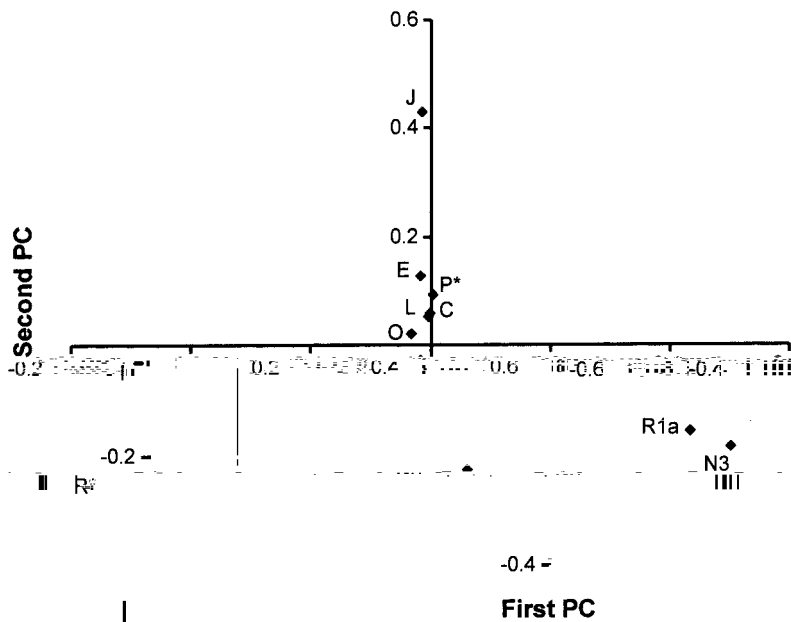


Figure 5 Plot of the contribution of each haplogroup to the first and second PC in the analysis of figure 4

groups). A finer haplogrouping, however, requires larger sample sizes to yield robust results.

The visualization of high-dimensional data by means of projection should not, in itself, serve as an inference tool. Our PC analysis does, however, suggest targets for further study. For instance, the grouping together of central and eastern Mediterranean populations spotlights the role of gene flow along the Mediterranean at various times. PC analysis, even of a single-marker system such as mtDNA or the Y chromosome, therefore has a part

- the human Y chromosome, disease and selection. *Trends Genet* 16:356–362
- Lutz S, Wessier H-J, Heizmann J, Pollak S (1998) Location and frequency of polymorphic positions in the mtDNA control region of individuals from Germany. *Int J Leg Med* 111:67–77
- Macaulay V, Richards M, Hickey E, Vega E, Cruciani F, Guida V, Scozzari R, Bonn -Tamir B, Sykes B, Torroni A (1999) The emerging tree of west Eurasian mtDNAs: a synthesis of control-region sequences and RFLPs. *Am J Hum Genet* 64:232–249
- Menozi P, Piazza PA, Cavalli-Sforza LL (1978) Synthetic maps of human gene frequencies in Europeans. *Science* 201:786–792
- Pfeiffer H, Brinkmann B, Huhne J, Rolf B, Morris AA, Steighner R, Holland MM, Forster P (1999) Expanding the forensic German mitochondrial DNA control region database: genetic diversity as a function of sample size and microgeography. *Int J Legal Med* 112:291–298
- Price TD (2000) *Europe's first farmers*. Cambridge University Press, Cambridge
- Richards M, Macaulay V (2000) Genetic data and the colonization of Europe: genealogies and founders. In: Renfrew C, Boyle K (eds) *Archaeogenetics: DNA and the population prehistory of Europe*. McDonald Institute for Archaeological Research, Cambridge, pp 139–151
- Richards MB, Macaulay VA, Bandelt H-J, Sykes BC (1998) Phylogeography of mitochondrial DNA in western Europe. *Ann Hum Genet* 62:241–260
- Richards M, Macaulay V, Hickey E, Vega E, Sykes B, Guida V, Rengo C, et al (2000) Tracing European founder lineages in the Near Eastern mitochondrial gene pool. *Am J Hum Genet* 67:1251–1276
- Rosser ZH, Zerjal T, Hurles ME, Adojaan M, Alavantic D, Amorim A, Amos W, et al (2000) Y-chromosomal diversity in Europe is clinal and influenced primarily by geography, rather than language. *Am J Hum Genet* 67:1526–1543
- Semino O, Passarino G, Brega A, Fellous M, Santachiara-Benerecetti S (1996) A view of the Neolithic demic diffusion in Europe through two Y chromosome-specific markers. *Am J Hum Genet* 59:964–968
- Semino O, Passarino G, Oefner PF, Lin AA, Arbuzova S, Beckman LE, De Benedictis G, Francalacci P, Kouvatisu A, Limborska S, Marcikiae M, Mika A, Mika B, Primorac D, Santachiara-Benerecetti S, Cavalli-Sforza LL, Underhill PA (2000) The genetic legacy of Paleolithic *Homo sapiens sapiens* in extant Europeans: a Y chromosome perspective. *Science* 290:1155–1159
- Simoni L, Calafell F, Pettener D, Bertranpetit J, Barbujani B