Unraveling male and female histories from human genetic data

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The increasing availability of large-scale genetic datasets has made it possible to ask detailed questions about the structure of human genetic diversity, and what that structure can teach us about human demographic history. Global, multi-locus analyses have suggested that human genetic diversity may fall into clusters that correspond approximately to continental origin. Detailed comparisons of mitochondrial DNA and the Y chromosome have revealed a history of sex-biased migration patterns that can vary widely across human populations. These patterns can be understood, however, when we incorporate our knowledge of local histories and cultural practices into our genetic analyses.

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Current Opinion in Genetics & Development 2006, **16**:611–617

This review comes from a themed issue on
Genomes and evolution
Edited by Chris Tyler-Smith and Molly Przeworski

Available online 25th October 2006

0959-437X/$ – see front matter

DOI 10.1016/j.gde.2006.10.004

Introduction

Human genetic data has become a valuable source of information about our past: where we came from, when we first arrived in different regions, and how our patterns of reproduction and migration have changed over time. The two chromosomes for which we have the most data are the mitochondrial chromosome (mtDNA; see Glossary) and the non-recombining portion of the Y chromosome (NRY; see Glossary). These chromosomes have unusual features that distinguish them from the rest of the genome: they are large, non-recombining, and uniparentally inherited. The strengths and limitations of inferences based on the NRY and mtDNA both rest on the fact that each of them is, in effect, a single genetic locus. The lack of recombination makes it possible to reconstruct the genealogy of each of these chromosomes with relatively high precision. However, the genealogical process is a highly variable one, and any single genealogy provides only limited insight into the underlying demographic processes that created it.

Large-scale molecular datasets have recently started to become available for autosomal and X-linked loci. Individually, each of these loci provides less information than either the NRY or mtDNA data. On the autosomes and X chromosome, our ability to infer the genealogical history of any particular chromosomal region is limited by the rate of recombination. Taken together, however, these data might eventually provide much greater statistical power for testing detailed hypotheses about human demographic history.

The uniparental inheritance of the NRY and mtDNA means that patterns of diversity observed for these chromosomes reflect different aspects of human history. The NRY, which is passed from father to son, reflects the demographic history of males, whereas the maternally inherited mtDNA reflects that of females. In principle, comparisons of the patterns of diversity found on these two chromosomes can reveal systematic differences between male and female patterns of reproduction and migration.

In the first half of this review, I discuss some of the recent studies that have used large, multi-locus datasets to assess the structure of human genetic diversity. The second half discusses genetic analyses of sex-biased demographic history, and focuses particularly on recent studies that have made direct comparisons between mtDNA and NRY samples drawn from the same populations.

Partitioning human genetic diversity

One common approach to the analysis of genetic data is to calculate various F-statistics (e.g. F_{ST} [see Glossary]), which describe the degree of genetic divergence among populations relative to the degree of genetic diversity within populations. For instance, F_{ST} describes just how much more genetically similar two individuals are likely to be if they come from the same subpopulation, compared with the genetic similarity of two random individuals. Invoking simple models of geographic structure, these estimates can be converted into migration rate estimates. For instance, under the symmetric island model of migration (see Glossary), the expected value of F_{ST} is approximately $1/(1 + 4Nm)$ (for autosomal loci; for the NRY and mtDNA, F_{ST} is approximately $1/(1 + Nm)$), where N is the population size of each subpopulation, or deme, and m is the probability of migration from one deme to another [1]. Within models of isolation by distance [2], the quantity $F_{ST}/(1 - F_{ST})$ calculated for pairs of populations is expected to increase with the geographic distance separating those populations. The dispersal rate can be estimated from this rate of increase [3].

A second approach, which has become popular in recent years, is the application of clustering techniques

Glossary

Effective population size: Roughly speaking, this is the effective number of individuals that contribute reproductively to the next generation. The effective population size is affected by a number of factors, including the census population size, and the variance of reproductive success. Specifically, the more skewed the distribution of reproductive success is in a population, the smaller its effective population size will be relative to its census size.

F_{ST}: This is one of a large group of closely related summary statistics used to characterize the degree of genetic/geographic structure in a population. Roughly, it can be thought of as the increased likelihood that two individuals (or genes) are genetically identical (or similar) when they are sampled from the same subpopulation, as opposed to being sampled at random from the entire population.

mtDNA: Mitochondrial DNA. In humans, mtDNA is maternally inherited, and its diversity is shaped by female demographic history.

NRY: The non-recombining portion of the Y chromosome. The NRY is paternally inherited and reflects male demographic history.

Symmetric island model of migration: A simple model of population structure in which the population is divided into a number of subpopulations, or demes. Each deme has a population of size N and exchanges migrants with other demes at a rate m. A migrant is equally likely to come from any other deme in the population. The product Nm is the average number of migrants that a deme receives each generation, and determines the degree to which the population is genetically structured.

to genetic data. The tool most commonly used for assessing human diversity in this context is the program STRUCTURE [4,5], which assigns individuals, or individual alleles, to a predefined number of clusters on the basis of genetic similarity.

When compared with other primates, human genetic diversity is surprisingly small. All contemporary human mtDNA chromosomes share a common ancestor approximately 200 thousand years ago (kya) [6–8]. The common ancestor of the NRY is even more recent, with estimates ranging from 46 to 109 kya [7–11]. This recent common ancestry is thought to reflect a recent African origin of anatomically modern humans with little or no genetic contribution from the populations who had left Africa much earlier (see Hammer *et al.* [12] and Reed and Tishkoff [13], this issue).

Statistical analyses have indicated a lack of genetic differentiation among the world's populations. Early studies, based on protein polymorphisms, found that the genetic differences among continents were small compared with the variation found within groups [14]. As more detailed molecular data has become available, analogous calculations have provided similar results: on average, two people from different continents differ genetically from each other only slightly more than two people from the same group.

These results have typically been interpreted as suggesting that there is no genetic basis for categorizing humans into discrete sub-types, or races. More recent results, based on the clustering algorithm employed by STRUCTURE, have seemed to contradict this conclusion. When this latter method was applied to autosomal microsatellite data from the Human Genome Diversity Panel [15], individuals clustered robustly by continental origin [16]. Qualitatively similar results were subsequently found when STRUCTURE was applied to X chromosome data [17••], and data from *Helicobacter pylori*, which colonizes the human stomach lining [18–20]. Although these studies have also found the degree of genetic differentiation among the continents to be small compared with the within-group variance, they do suggest that human genetic diversity is somewhat multi-modal, and that categorization into discrete groups might have some basis.

Although the clustering by continental origin observed in this genetic data appears to be well established, the cause of that clustering is a matter of debate. It has been suggested that the clustering is simply an artefact of the sampling scheme: the sites from which data were collected were clustered, and the underlying genetic diversity might reveal a smoother cline [21•]. Another possibility is that the apparent discontinuities in the genetic diversity reflect the presence of large-scale geographic barriers (e.g. oceans, mountains and deserts) that reduce gene flow at this largest geographic scale [22•]. This latter view is further supported by recent studies that have successfully explained more of the features of human genetic diversity by incorporating more realistic geographic constraints to human gene flow [23•,24•].

Marital residence and diffusive migration

Collections of mtDNA and NRY have been used to compare male and female demographic patterns (e.g. migration) across a variety of geographic and cultural scales. In this context, it is useful to distinguish two different types of migration. One, which I will call 'diffusive migration', consists of the uncorrelated movements of individuals or small groups. The other I will call 'directional migration', wherein larger groups move in roughly the same direction, possibly for multiple generations.

Diffusive migration can be characterized by the intergenerational migration distance — the average distance, in some sense, between the birthplaces of parents and offspring. The smaller this distance, the more geographic structure we expect to find in the genetic data. Several studies have invoked marital residence to explain differences found in the degree of geographic structure evident on the mtDNA and NRY. For instance, genetic patterns consistent with patrilocality have been identified in the Caucasus [25], in Russia [26], and among populations of Kurds [27] and Kalmyks [28]. Table 1 presents results from a few of the recent studies that compare mtDNA and NRY diversity, focusing particularly on those studies in which the two chromosomes have been sampled from the same sets of populations.

Table 1

Estimates of sex-biased migration based on partitioning of genetic variance.

Region	mtDNA F_{ST}	NRY F_{ST}	Male:female *Nm*	NRY Data	Notes	References
Global	0.186	0.645	0.13	STR		[49]
Global	0.401	0.357	1.21	Seq	Overall	[50••]
	0.261	0.209	1.34		Within continents	
	0.189	0.187	1.01		Among continents	
Thailand	0.290	0.131	2.71	STR	Matrilocal	[29]
	0.118	0.450	0.16		Patrilocal	
	0.038	0.130	0.26		Food-producers	
Sub-Saharan Africa	0.431	0.072	9.76	SNP	Hunter-gatherer	[31••]
	0.025	0.174	0.12		Overall	
Caucasus	0.025	0.174	0.12	SNP	Overall	[25]
	0.008	0.060	0.13		Within groups	
	0.018	0.121	0.13		Among groups	
Sub-Saharan Africa	0.16	0.33	0.39	SNP	Overall	[40••]
	0.13	0.28	0.38		Within groups	
	0.04	0.06	0.65		Among groups	

This table lists the reported F_{ST} values for mtDNA and NRY data from a number of recent studies, with the geographic region studied listed in the first column. With the exception of the first study, by Seilstad *et al* [49], each of these studies is based on mtDNA and NRY samples drawn from the same individuals. The male:female *Nm* ratio was calculated assuming that $Nm = (1/F_{ST})–1$. Ratio values greater than 1 correspond to higher male migration (i.e. higher male effective population size), whereas ratios less than 1 correspond to higher female migration (i.e. higher female effective population size). The NRY data column indicates what type of data was collected from the Y chromosome: Seq, direct sequencing; SNP, single nucleotide polymorphisms; STR, microsatellite repeats. The mtDNA was all collected by direct sequencing. For each study in which the authors performed analyses on different subsets of the data, or at different levels of resolution, the estimated *Nm* ratio for each sub-analysis is shown, with the partitioning of the data indicated in the Notes column.

In each case, the authors' estimates of F_{ST} for the two genetic systems are shown. In some cases, the partitioning of genetic diversity was calculated at different scales, or for different subsets of the data. For each pair of estimated F_{ST} values, I have calculated the ratio of the corresponding estimates of *Nm* for males and females. If we assume that the male and female effective population sizes (see Glossary) are similar (or, equivalently, that the differences between male and female reproductive skew are negligible), this ratio provides a crude measure of the relative magnitudes of male and female migration, with values less than one corresponding to female-biased migration (patrilocality) and values greater than one corresponding to male-biased migration (matrilocality). We will return to the question of just how similar the male and female effective population sizes are in a later section.

The most striking feature of the data in Table 1 is the magnitude of the variation in the male–female migration ratio across different regions and cultures. This ratio ranges from 0.12 (~8-fold higher female migration) in the Caucasus to 9.76 (~10-fold higher male migration) among sub-Saharan forager populations. Studies that have focused on particular geographic regions and/or groups of populations have found genetic patterns that correspond with known cultural traits. For instance, the migration ratio reflects ethnographically defined patterns of marital residence in the Thai hill tribes [29,30] and correlates with the mode of subsistence (i.e. hunting and gathering as opposed to food-producing) in Sub-Saharan Africa [31••].

The fact that genetic diversity is heavily influenced by cultural practices, particularly at local scales, emphasizes the importance of analytic methods that can account for the heterogeneity — both across cultures and through time — of human demographic processes. For contemporary populations, we can observe this heterogeneity directly. For ancient populations, we can partially reconstruct this heterogeneity on the basis of patterns observed in the ethnographic and archaeological data. For instance, food-producing (i.e. pastoralist and agricultural) populations are predominantly patrilocal, but contemporary foragers exhibit a broad mixture of marital residence [32]. This difference can be seen in comparisons of contemporary foraging and food-producing populations [31••], but also has implications for the history of marital residence patterns on a global scale. The transition to a food-producing lifestyle was probably accompanied by an increase in patrilocality. The genetic consequences of this transition might still be reflected in the patterns of mtDNA and NRY diversity at different geographic scales [33].

Directional migration, range expansion, and hypergamy

The current standard model has anatomically modern humans arising in Africa ~100–200 kya, and only recently

expanding to the rest of the world. These migrations would have consisted of groups of men and women moving together, and on a global scale, the genetic evidence from the mtDNA and NRY are qualitatively similar. On more local scales, human genetic diversity has also been influenced by the more recent expansion of particular populations. Some of these events have left similar genetic signatures on the mtDNA and NRY, suggesting that these movements involved similar numbers of men and women, such as the Mongolian origins of the Kalmyks [28], and the African–Indonesian admixture giving rise to the indigenous peoples of Madagascar [34].

Other migrations have consisted primarily of men, who then admix with the pre-existing population. The result is an introgression of NRY, but not mtDNA, from the immigrants into the aboriginal population. Studies of native South American populations have revealed high frequencies of European-typical Y chromosomes, but not mtDNA [35–37]. The introgression of European NRY haplotypes has also been documented in the Inuit of Greenland [38] (see Table 2). Other cases of asymmetric gene flow have also been associated with known historical events, such as warfare and commerce, that predominantly involved the movements of males. On the Mediterranean island of Ibiza, mtDNA patterns appear to reflect genetic isolation of the Carthaginian–Phoenician founder population. NRY diversity on this island, however, shows evidence of repeated contact with mainland Spain, Italy and North Africa [39]. Genetic analysis of the Caucasus indicates a genetic affinity for West Asian populations specifically on the NRY, suggesting a history of male-dominated contact with those regions [25].

Other asymmetries in male and female demography might have been driven by particular cultural innovations, such as the domestication of crops and animals. Historically, agricultural populations have expanded at the expense of neighboring forager populations. Contact between the forager and food-producing populations often involves hypergamy, in which forager females marry food-producing males and are assimilated into the expanding agricultural community. The analogous assimilation of males is far less common. Genetic patterns consistent with a history of agricultural expansion and hypergamy have been characterized in Sub-Saharan Africa [31••,40••]. In this case, Bantu-speaking food producers have expanded from a West-African homeland, assimilating mtDNA lineages from the neighboring forager populations. A similar asymmetry can be seen in the movement of Tibeto-Burman populations into Southeast Asia [41].

Effective population size

Male and female demographies can differ not only in their patterns of migration, but in their effective population sizes. In most mammals, males have a higher variance of reproductive success than females, leading to reduced genetic diversity. Comparisons of the time to the most recent common ancestor (TMRCA) for mtDNA and NRY genealogies suggest that this holds true for humans as well, with the effective population size of males being roughly half that of females [8].

However, this global value represents some average effective size over time and across cultures. As with migration patterns, the distribution of human reproduc-

Table 2

Sex-specific admixture estimates.

Region	Genetic Source	mtDNA	NRY	References
Sub-Saharan Africa	European	0%	12%	[51]
	African	100%	87%	
South America	Amerindian	95–100%	35–97%	[35]
	African	0–5%	0–11%	
	European	0–5%	3–60%	
South America	Amerindian	90%	1%	[37]
	African	0–5%	4%	
	European	5–10%	95%	
Greenland	European	0%	58%	[38]
	Amerindian	100%	42%	
Southeast Asia	Northeast Asia	44%	62%	[41]
	Southeast Asia	56%	38%	
Madagascar	African	38%	51%	[34]
	Indonesian	62%	49%	

This table summarizes the conclusions of a number of studies focusing on particular populations known to have undergone admixture in the past, with the locations of these populations listed in the first column. The source populations that contributed to the admixed population are listed in the second column, and their relative contributions are listed in the third and fourth columns. The third column lists the maternal contribution of each source population to the admixed population — estimated from mtDNA data. The fourth column lists the corresponding paternal contributions — estimated from NRY data.

Table 3

Ethnographic and archaeological data relevant to sex-specific demography

Observation				References
Human generation interval (years)		Male	Female	[52]
Developed nations		30.8	27.3	
Less-developed nations		31.8	28.3	
Forager populations		31.5	25.6	
Residential floor area (patrilocal)		14.5–42.7 m^2		[53]
Residential floor area (matrilocal)		79.2–270.8 m^2		
	Marital residence			
	Patrilocal	Multilocal	Matrilocal	[32]
Non-Foragers	60%	25.3%	14.7%	
Foragers	34.3%	42.9%	22.9%	

This table lists values from ethnographic and archaeological data that might be useful for calibrating future genetic studies of sex-biased demographic processes in humans. The first set of entries lists the average generation time for males and females in different types of populations. These values are needed to translate between genetic time (generations) and calendar time (years). The second set of entries refers to the differences in the sizes of individual residences in matrilocal and patrilocal societies. This type of data is useful for reconstructing the spatio-temporal pattern of sex-biased human migration. The final set of values describes the distribution of marital residence patterns among populations in the ethnographic record. These values suggest that a shift to patrilocality co-occurs with the transition to agriculture.

tive success is sensitive to cultural practices and can change rapidly. Cultural practices related to the relative male and female effective population sizes, such as degree of polygyny, vary systematically with features such as mode of subsistence, latitude and state structure [42,43]. For instance, natives of New Guinea have dramatically reduced genetic diversity on the NRY relative to the mtDNA [44]. This reduction is probably due in part to high rates of polygyny — in some communities, ~30% of men have more than one wife, and ~40% are unmarried — resulting in a large male reproductive skew. Warfare has also been frequent among these societies, with high male (but low female) mortality rates.

Although we expect most human cultures to have a lower male than female effective population size, particular cultural practices can locally produce the opposite pattern. Genetic analysis of the Samaritan populations have revealed the persistence of four distinct NRY haplotypes, despite strict endogamy and a dramatic reduction in population size in the twentieth century (dropping from more than one million in late Roman times to only 146 individuals in 1917; even today, the population numbers only 640) [45]. In this case, the presence of strict marriage rules among four distinct families has guaranteed the persistence of these four paternal lineages, despite this population bottleneck.

What next?

Humans are unusual in the extent to which our demographic and genetic patterns have been shaped by cultural practices. For many other species, it may be reasonable to assume that the patterns of reproduction and migration we observe today are similar to those that existed in the past. For humans, we know that these patterns vary across different geographic regions and different cultures. We also know that these patterns have changed over time, as innovations from agriculture to air travel have continually reshaped the way that people (and their genes) move around the world.

In addition to the intrinsic complexity of human demographic history, issues of sampling and ascertainment bias hampered many of the early efforts in this area. Several authors have recently discussed these issues in the particular context of human genetics [21•,22•,32,46–48]. Fortunately, many recent study designs have taken these issues more seriously, resulting in higher-quality datasets and more reliable inferences.

Taking full advantage of the information in the patterns of human genetic diversity will require the development of more complex and realistic models. These models will have to incorporate geographic, linguistic, archaeological and ethnographic data (see Table 3 for a few examples). Some of the studies mentioned here have already begun to integrate this type of complexity into their analyses, with exciting results. As this field moves into the future, we will try to draw more detailed inferences from genetic data, and the questions of how to collect and analyze genetic data in a meaningful way will continually have to be revisited.

References and recommended reading

Papers of particular interest, published within the annual period of review, have been highlighted as:

- • of special interest
- •• of outstanding interest

1. Wright S: **The genetical structure of populations**. *Ann Eugen* 1950, **15**:323-354.

2. Wright S: **Isolation by distance**. *Genetics* 1943, **28**:114-138.

3. Rousset F: **Genetic differentiation and estimation of gene flow from F-statistics under isolation by distance**. *Genetics* 1997, **145**:1219-1228.

4. Pritchard JK, Stephens M, Donnelly P: **Inference of population structure using multilocus genotype data**. *Genetics* 2000, **155**:945-959.

5. Falush D, Stephens M, Pritchard JK: **Inference of population structure using mutilocus genotype data: Linked loci and correlated allele frequencies**. *Genetics* 2003, **164**:1567-1587.

6. Ingman M, Kaessmann H, Paabo S, Gyllensten U: **Mitochondrial genome variation and the origin of modern humans**. *Nature* 2000, **408**:708-713.

7. Tang H, Siegmund DO, Shen PD, Oefner PJ, Feldman MW: **Frequentist estimation of coalescence times from nucleotide sequence data using a tree-based partition**. *Genetics* 2002, **161**:447-459.

8. Wilder JA, Mobasher Z, Hammer MF: **Genetic evidence for unequal effective population sizes of human females and males**. *Mol Biol Evol* 2004, **21**:2047-2057.

9. Pritchard JK, Seielstad MT, Perez-Lezaun A, Feldman MW: **Population growth of human Y chromosomes: a study of Y chromosome microsatellites**. *Mol Biol Evol* 1999, **16**:1791-1798.

10. Thomson R, Pritchard JK, Shen PD, Oefner PJ, Feldman MW: **Recent common ancestry of human Y chromosomes: Evidence from DNA sequence data**. *Proc Natl Acad Sci USA* 2000, **97**:7360-7365.

11. Hammer MF, Zegura SL: **The human Y chromosome haplogroup tree: nomenclature and phylogeography of its major divisions**. *Annu Rev Anthropol* 2002, **31**:303-321.

12. Wall JD, Hammer MF: **Archaic admixture in the human genome.** *Curr Opin Genet Dev* 2006, in press.

13. Tishkoff S: **African human diversity, origins and migrations.** *Curr Opin Genet Dev* 2006, in press.

14. Lewontin RC: **The apportionment of human genetic diversity**. *Evolutionary Biology* 1972, **6**:381-398.

15. Cann HM, de Toma C, Cazes L, Legrand MF, Morel V, Piouffre L, Bodmer J, Bodmer WF, Bonne-Tamir B, Cambon-Thomsen A *et al.*: **A human genome diversity cell line panel**. *Science* 2002, **296**:261-262.

16. Rosenberg NA, Pritchard JK, Weber JL, Cann HM, Kidd KK, Zhivotovsky LA, Feldman MW: **Genetic structure of human populations**. *Science* 2002, **298**:2381-2385.

17. •• Ramachandran S, Rosenberg NA, Zhivotovsky LA, Feldman MW: **Robustness of the inference of human population structure: a comparison of X-chromosomal and autosomal microsatellites**. *Human Genomics* 2004, **1**:87-97.

A multi-locus analysis of sex-biased demographic processes on a global scale. This type of analysis, based on the comparison of autosomal and X-linked markers, has the potential of providing more statistical power than mtDNA–NRY comparisons.

18. Wirth T, Wang XY, Linz B, Novick RP, Lum JK, Blaser M, Morelli G, Falush D, Achtman M: **Distinguishing human ethnic groups by means of sequences from *Heliclobacter pylori*: lessons from Ladakh**. *Proc Natl Acad Sci USA* 2004, **101**:4746-4751.

19. Falush D, Wirth T, Linz B, Pritchard JK, Stephens M, Kidd M, Blaser MJ, Graham DY, Vacher S, Perez-Perez GI *et al.*: **Traces of human migrations in *Helicobacter pylori* populations**. *Science* 2003, **299**:1582-1585.

20. Falush D, Kraft C, Taylor NS, Correa P, Fox JG, Achtman M, Suerbaum S: **Recombination and mutation during long-term gastric colonization by *Helicobacter pylori*: estimates of clock rates, recombination size, and minimal age**. *Proc Natl Acad Sci USA* 2001, **98**:15056-15061.

21. • Serre D, Paabo SP: **Evidence for gradients of human genetic diversity within and among continents**. *Genome Res* 2004, **14**:1679-1685.

See annotation [22•].

22. • Rosenberg NA, Mahajan S, Ramachandran S, Zhao C, Pritchard JK, Feldman MW: **Clines, clusters, and the effect of study design on the inference of human population structure**. *Plos Genet* 2005, **1**:660-671.

The studies by Serre *et al*. [21•] and Rosenberg *et al*. [22•] debate the impact of the geographical distribution of sampling sites on the inference of clusters of human genetic diversity.

23. • Ray N, Currat M, Berthier P, Excoffier L: **Recovering the geographic origin of early modern humans by realistic and spatially explicit simulations**. *Genome Res* 2005, **15**:1161-1167.

See annotation [24•].

24. • Ramachandran S, Deshpande O, Roseman CC, Rosenberg NA, Feldman MW, Cavalli-Sforza LL: **Support from the relationship of genetic and geographic distance in human populations for a serial founder effect originating in Africa**. *Proc Natl Acad Sci USA* 2005, **102**:15942-15947.

The studies by Ray *et al*. [23•] and Ramachandran *et al*. [24•] incorporate more realistic models of geographic structure into the analysis of global patterns of genetic diversity.

25. Nasidze I, Ling EYS, Quinque D, Dupanloup I, Cordaux R, Rychkov S, Naumova O, Zhukova O, Sarraf-Zadegan N, Naderi GA *et al.*: **Mitochondrial DNA and Y-chromosome variation in the Caucasus**. *Ann Hum Genet* 2004, **68**:205-221.

26. Malyarchuk B, Derenko M, Grzybowski T, Lunkina A, Czarny J, Rychkov S, Morozava I, Denisova G, Miscicka-Sliwka D: **Differentiation of mitochondrial DNA and Y chromosomes in Russian populations**. *Hum Biol* 2004, **76**:877-900.

27. Nasidze I, Quinque D, Ozturk M, Bendukidze N, Stoneking AM: **MtDNA and Y-chromosome variation in Kurdish groups**. *Ann Hum Genet* 2005, **69**:401-412.

28. Nasidze I, Quinque D, Dupanloup I, Cordaux R, Kokshunova L, Stoneking M: **Genetic evidence for the Mongolian ancestry of Kalmyks**. *Am J Phys Anthropol* 2005, **128**:846-854.

29. Oota H, Settheetham-Ishida W, Tiwawech D, Ishida T, Stoneking M: **Human mtDNA and Y-chromosome variation is correlated with matrilocal versus patrilocal residence**. *Nat Genet* 2001, **29**:20-21.

30. Hamilton G, Stoneking M, Excoffier L: **Molecular analysis reveals tighter social regulation of immigration in patrilocal populations than in matrilocal populations**. *Proc Natl Acad Sci USA* 2005, **102**:7476-7480.

31. •• Destro-Bisol G, Donati F, Coia V, Boschi I, Verginelli F, Caglia A, Tofanelli S, Spedini G, Capelli C: **Variation of female and male lineages in sub-Saharan populations: the importance of sociocultural factors**. *Mol Biol Evol* 2004, **21**:1673-1682.

This study incorporates linguistic and ethnographic information into the analysis of genetic data to study the interplay between culture and demography. See also the study by Wood *et al*. [40••].

32. Marlowe FW: **Marital residence among foragers**. *Curr Anthropol* 2004, **45**:277-284.

33. Wilkins JF, Marlowe FW: **Sex-biased migration in humans: what should we expect from genetic data?** *Bioessays* 2006, **28**:290-300.

34. Hurles ME, Sykes BC, Jobling MA, Forster P: **The dual origin of the Malagasy in Island Southeast Asia and East Africa: evidence from maternal and paternal lineages**. *Am J Hum Genet* 2005, **76**:894-901.

35. Mesa NR, Mondragon MC, Soto ID, Parra MV, Duque C, Ortiz-Barrientos D, Garcia LF, Velez ID, Bravo ML, Munera JG *et al.*: **Autosomal, mtDNA, and Y-chromosome diversity in Amerinds: pre- and post-Columbian patterns of gene flow in South America**. *Am J Hum Genet* 2000, **67**:1277-1286.

36. Carvajal-Carmona LG, Ophoff R, Service S, Hartiala J, Molina J, Leon P, Ospina J, Bedoya G, Freimer N, Ruiz-Linares A: **Genetic demography of Antioquia (Colombia) and the Central Valley of Costa Rica**. *Hum Genet* 2003, **112**:534-541.

37. Carvajal-Carmona LG, Soto ID, Pineda N, Ortiz-Barrientos D, Duque C, Ospina-Duque J, McCarthy M, Montoya P, Alvarez VM, Bedoya G *et al.*: **Strong Amerind/white sex bias and a possible sephardic contribution among the founders of a population in northwest Colombia**. *Am J Hum Genet* 2000, **67**:1287-1295.

38. Bosch E, Calafell F, Rosser ZH, Norby S, Lynnerup N, Hurles ME, Jobling MA: **High level of male-biased Scandinavian admixture**

in Greenlandic Inuit shown by Y-chromosomal analysis. *Hum Genet* 2003, **112**:353-363.

39. Tomas C, Jimenez G, Picornell A, Castro JA, Ramon MM: **Differential maternal and paternal contributions to the genetic pool of Ibiza island, Balearic archipelago**. *Am J Phys Anthropol* 2006, **129**:268-278.

40. •• Wood ET, Stover DA, Ehret C, Destro-Bisol GD, Spedini G, McLeod H, Louie L, Bamshad M, Strassmann BI, Soodyall H *et al.*: **Contrasting patterns of Y chromosome and mtDNA variation in Africa: evidence for sex-biased demographic processes**. *Eur J Hum Genet* 2005, **13**:867-876.
This study incorporates linguistic and ethnographic information into the analysis of genetic data to study the interplay between culture and demography. See also the study by Destro-Bisol *et al*. [31••].

41. Wen B, Xie XH, Gao S, Li H, Shi H, Song XF, Qian TZ, Xiao CJ, Jin JZ, Su B *et al.*: **Analyses of genetic structure of Tibeto-Burman populations reveals sex-biased admixture in southern Tibeto-Burmans**. *Am J Hum Genet* 2004, **74**:856-865.

42. Marlowe FW: **The mating system of foragers in the standard cross-cultural sample**. *Cross-Cultural Res* 2003, **37**:282-306.

43. Marlowe FW: **Hunter-gatherers and human evolution**. *Evol Anthropol* 2005, **14**:54-67.

44. Kayser M, Brauer S, Weiss G, Schiefenhovel W, Underhill P, Shen PD, Oefner P, Tommaseo-Ponzetta M, Stoneking M: **Reduced Y-chromosome, but not mitochondrial DNA, diversity in human populations from West New Guinea**. *Am J Hum Genet* 2003, **72**:281-302.

45. Shen P, Lavi T, Kivisild T, Chou V, Sengun D, Gefel D, Shpirer I, Woolf E, Hillel J, Feldman MW *et al.*: **Reconstruction of patrilineages and matrilineages of Samaritans and other Israeli populations from Y-chromosome and mitochondrial DNA sequence variation**. *Hum Mutat* 2004, **24**:248-260.

46. Ptak SE, Przeworski M: **Evidence for population growth in humans is confounded by fine-scale population structure**. *Trends Genet* 2002, **18**:559-563.

47. Hammer MF, Blackmer F, Garrigan D, Nachman MW, Wilder JA: **Human population structure and its effects on sampling Y chromosome sequence variation**. *Genetics* 2003, **164**:1495-1509.

48. Clark AG, Hubisz MJ, Bustamante CD, Williamson SH, Nielsen R: **Ascertainment bias in studies of human genome-wide polymorphism**. *Genome Res* 2005, **15**:1496-1502.

49. Seielstad MT, Minch E, Cavalli-Sforza LL: **Genetic evidence for a higher female migration rate in humans**. *Nat Genet* 1998, **20**:278-280.

50. •• Wilder JA, Kingan SB, Mobasher Z, Pilkington MM, Hammer MF: **Global patterns of human mitochondrial DNA and Y-chromosome structure are not influenced by higher migration rates of females versus males**. *Nat Genet* 2004, **36**:1122-1125.
A global analysis based on resequenced Y chromosome data, which avoids many of the ascertainment bias problems associated with other Y-chromosome analyses.

51. Beleza S, Gusmao L, Amorim A, Carracedo A, Salas A: **The genetic legacy of western Bantu migrations**. *Hum Genet* 2005, **117**:366-375.

52. Fenner JN: **Cross-cultural estimation of the human generation interval for use in genetics-based population divergence studies**. *Am J Phys Anthropol* 2005, **128**:415-423.

53. Peregrine PN: **Cross-cultural approaches in archaeology: comparative ethnology, comparative archaeology, and archaeoethnology**. *J Archaeol Res* 2004, **12**:281-309.