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# Y-chromosomal DNA variation and human population history

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The Y chromosome has unique genetic properties that make it particularly useful for some forensic and evolutionary purposes. It is haploid, male-specific and escapes recombination over most of its length, so it changes only by the gradual accumulation of mutations in distinct lineages. Its effective population size is one quarter of that of autosomes and the genetic mobility of males may be lower than that of females in many societies. The Y is therefore greatly affected by genetic drift and shows very high levels of geographical differentiation. More than 200 slowly evolving binary markers (mainly SNPs) and over 30 rapidly evolving STRs are now available, and combinations of these can be highly informative.

The common patterns of Y-chromosomal variation are illustrated by studies of haplogroup distribution in Europe and STR variation in Pakistan. A collaborative study of 3616 men from 47 populations in Europe and neighbouring countries was carried out using 11 binary markers known to be informative in this area (Rosser et al. (2000) *Am. J. Hum. Genet.* **67**, 1526–1543). Six common haplogroups were found and five of these showed clinal patterns of variation, which were probably established by the entry of modern humans in the Palaeolithic and multiple but minor subsequent migrations. Large-scale STR analyses of these European samples are not yet available, but 711 men from 12

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populations in Pakistan have been analysed with 16 STRs (Mohyuddin et al. (2001) *Forensic Sci. Int.* **118**, 141–146). Most haplotypes were found in single individuals and only 2 were present in more than 10 men, in each case largely or entirely from the same population.

We have now analysed 1274 men from Central Asia, China and nearby countries with 16 binary markers and 16 STRs, and find significantly different patterns of Y variation. Large differences in haplogroup frequency are often seen between neighbouring populations, suggesting a high degree of genetic isolation and significant bottlenecks or founder effects. Single lineages with low levels of STR variation can be present at high frequency in individual populations. For example, different lineages in the Kazaks and Kyrgyz had coalescence times of $\sim$ 700 years (with large uncertainties). In addition, one lineage represented almost 10% of the chromosomes found in a wide area stretching from the Pacific to the Caspian Sea. This lineage had a coalescence time of $\sim$ 900 years. In contrast to the Kazak and Kyrgyz lineages, it was found in 16 different populations and has thus spread rapidly over a large region. Genealogical evidence from the Pakistani Hazara suggests that it may be that of Genghis Khan.