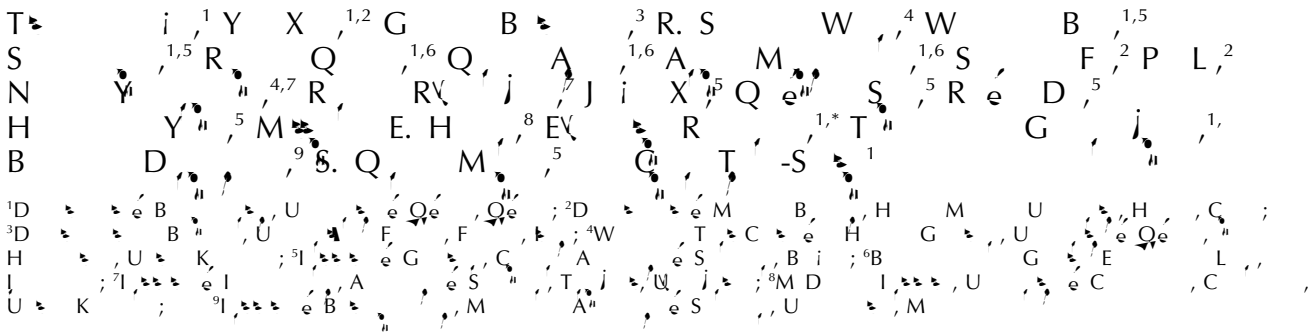


Report

The Genetic Legacy of the Mongols



We have identified a Y-chromosomal lineage with several unusual features. It was found in 16 populations throughout a large region of Asia, stretching from the Pacific to the Caspian Sea, and was present at high frequency: ~8% of the men in this region carry it, and it thus makes up ~0.5% of the world total. The pattern of variation within the lineage suggested that it originated in Mongolia ~1,000 years ago. Such a rapid spread cannot have occurred by chance; it must have been a result of selection. The lineage is carried by likely male-line descendants of Genghis Khan, and we therefore propose that it has spread by a novel form of social selection resulting from their behavior.

The patterns of variation found in human DNA are usually considered to result from a balance between neutral processes and natural selection. Among the former, mutation, recombination, and migration increase variation, whereas genetic drift decreases it. Natural selection can act to remove deleterious variants (purifying selection), maintain polymorphism (balancing selection), or produce a trend (directional selection). Clear examples of the latter are rare in humans, but probable cases, such as those associated with resistance to malaria (Hamblin and Di Rienzo 2000) or unidentified pathogens (Stephens et al. 1998), can be recognized by the “signature” they leave in the genome. The rapid increase in frequency of the selected allele and its linked sequences results in

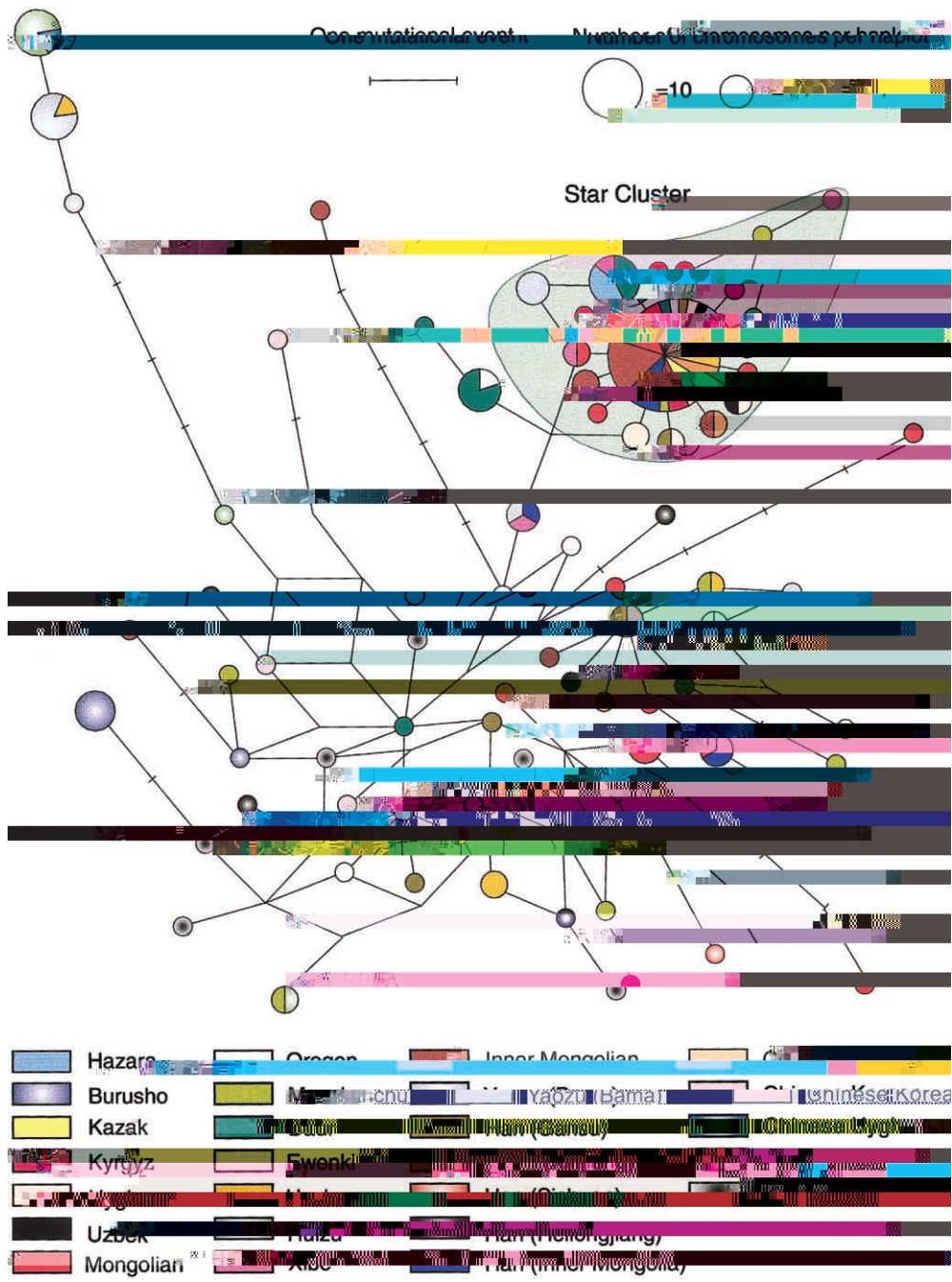


Figure 1

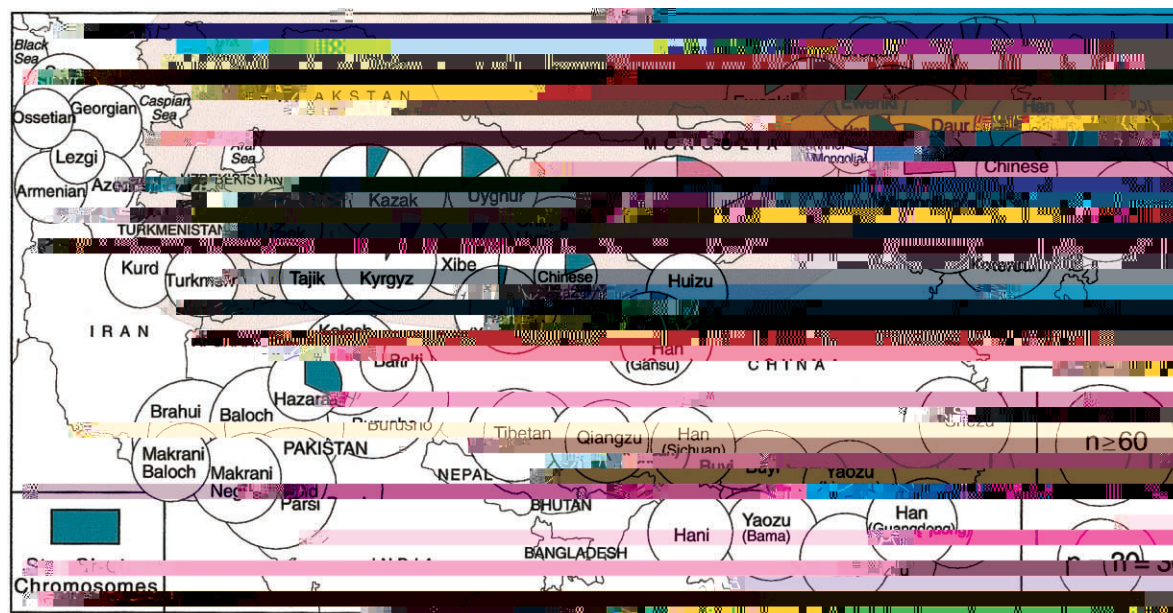


Figure 2 Geographical distribution of star-cluster chromosomes. Populations are shown as circles with an area proportional to sample size; star-cluster chromosomes are indicated by green sectors. The shaded area represents the extent of Genghis Khan’s empire at the time of his death (Morgan 1986).

We then used two approaches to calculate a TMRCA for the star-cluster chromosomes. The program BAT-WING (Wilson and Balding 1998) uses models of both mutation and population processes, which were specified as described elsewhere (Qamar et al. 2002). With this program, we estimated ~1,000 years for the TMRCA (95% confidence interval limits ~700–1,300 years). The use of alternative demographic models with constant or exponentially increasing population size changed the estimate by <10%. A method that does not consider population structure (Morral et al. 1994), ρ , suggested ~860 (~590–1,300) years. In both calculations, we assumed a generation time of 30 years. The origin was most likely in Mongolia, where the largest number of different star-cluster haplotypes is found (fig. 1). Thus, a single male line, probably originating in Mongolia, has spread in the last ~1,000 years to represent ~8% of the males in a region stretching from northeast China to Uzbekistan. If this spread were due to a general population expansion, we would expect to find multiple lineages with the same characteristics of high frequency and presence in multiple populations, but we do not (Zerjal et al. 2002). The star-cluster pattern is unique.

This rise in frequency, if spread evenly over ~34 generations, would require an average increase by a factor of ~1.36 per generation and is thus comparable to the most extreme selective events observed in natural populations, such as the spread of melanic moths in 19th-century England in response to industrial pollution

(Edleston 1865). We evaluated whether it could have occurred by chance. If the population growth rate is

close male relatives had many children. Although the Mongol empire soon disintegrated as a political unit, his male-line descendants ruled large areas of Asia for many generations. These included China, where the Yüan Dynasty emperors remained in power until 1368, after which the Mongols continued to dominate the co.776C

