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*Review*

# Use of molecular genetics and historical records to reconstruct the history of local communities

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**Recent advances in molecular genetics made the inference of past demographic events through the analysis of gene pools from modern populations possible. The technology uses genetic markers to provide previously unavailable resolution into questions of human evolution, migration and the historical relationship of separated human populations. Some of the genetic markers used to measure variation (polymorphism) within populations are found in the Y-chromosome and mitochondrial DNA. Variations in these two types of DNA can be grouped into continent-specific haplogroups or lineages. Geographic origin can be assigned to each lineage, and consequently trace back migration pattern of human populations. Y-chromosome and mitochondrial DNA are used to construct paternal and maternal lineages respectively. The use of these molecular techniques together with historical records in an integrated manner can greatly benefit the study of the social history of admixed communities, such as the Cape Muslim community living in the Western Cape of South Africa.**

**Key words:** Molecular genetics, genetic markers, Y-chromosome, and mitochondrial DNA.

## INTRODUCTION

The study of human evolution, migration and historical relationships of separated human populations requires a multidisciplinary approach. In addition to both historical and statistical research, various other disciplines such as physical and social anthropology, archaeology, demography and linguistics provide complementary approaches to researching questions regarding human evolution (Cavalli-Sforza and Feldman, 2003). Indeed, the advances in our understanding of the evolutionary history of humans attest to the advantages of this multidisciplinary research approach (Cavalli-Sforza and Feldman, 2003). Furthermore and with the advances in molecular genetic markers, it became possible to infer past demographic events through the analysis of gene pools from modern populations (Flores et al., 2003). Haploid markers from mitochondrial DNA and the Y chromosome, in particular, have proven invaluable for generating a standard model for evolution of modern humans (Cavalli-Sforza and Feldman, 2003).

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## IMPORTANCE OF HUMAN DNA VARIATIONS

Data on various kinds of DNA variation in human populations have rapidly accumulated, and there is increasing recognition of the importance of this variation for medicine and developmental biology, and for understanding the history of our species (Cavalli-Sforza and Feldman, 2003). By late twentieth century, population genetics research was marked by a significant expansion in the available research tools through a greater appreciation of the level of DNA variations in the human genome (Cavalli-Sforza and Feldman, 2003).

The analysis of these DNA variations or also called polymorphisms is making a valuable contribution to the understanding of human evolution (Jobling et al., 1998). It is often assumed that the polymorphisms observed at loci on mitochondrial DNA (mtDNA) and the Y chromosome are selectively neutral and, therefore, that existing patterns of molecular variation can be used to deduce the histories of populations in terms of drift, population movements, and cultural practices (Jobling et al., 1998).

The maternally inherited mitochondrial DNA and its paternally inherited counterpart, the Y chromosome have been widely used for the reconstruction of human history and dispersal of anatomically modern humans because of their uniqueness in the human genome (Peričić et al., 2005). mtDNA and the non-recombining region of Y (NRY) represent the only two haploid segments of human genome, since they are transmitted uniparentally, without reshuffling in each generation through the recombination process (Peričić et al., 2005). In fact, by escaping recombination, these two haploid systems are inherited as single loci that change only via mutations accumulating over time, thus allowing preservation of a relatively simple record of genetic history, in comparison to autosomal DNA (Peričić et al., 2005).

## INFERENCE OF POPULATION HISTORY FROM GENETIC DIVERSITY

Variations in the Y-chromosome and in its mtDNA counterpart can be grouped into continent-specific haplogroups or lineages (Y Chromosome Consortium, 2002; Jobling and Tyler-Smith, 2003). This geographic correlation of specific polymorphisms is helpful in uncovering past historic events and in explaining the genetic composition of extant populations (Gonçalves et al., 2003). Mitochondrial polymorphisms have been extensively used to study the maternal composition and relationships of human populations and past migrational events. Recently, Y-chromosomal markers have also acquired a special interest because they can provide an independent and paternal historic view of those relationships (Gonçalves et al., 2003).

For mtDNA, it was found that individuals from different geographic and ethnic origin have different restriction fragment patterns (Brown, 1980). Furthermore, combination of control and coding region data allows grouping of mtDNA variants into haplogroups defined by one or more mtDNA coding region polymorphisms and particular control region sequences. The majority of these haplogroups show geographic specificity, thus making mtDNA suitable for studies of maternal genetic history (Peričić et al., 2005).

For the Y chromosome on the other hand, a large number of Y chromosomal single nucleotide polymorphisms (SNP) were discovered since the implementation of denaturing high-performance liquid chromatography (Underhill et al., 1997). Combinations of slow-mutating and stable non-recurrent SNPs define Y chromosomal haplogroups, whereas highly-mutating recurrent short tandem repeat loci (STRs) define haplotypes. STRs, also called microsatellites, can be used to investigate demographic events that occurred in a more recent time-scale and, in combination with binary markers, they enable inferences to diversity (Peričić et al., 2005). Systems that combine SNPs and microsatellites may provide a way to map haplotypes more finely (Cavalli-Sforza and Feldman, 2003).

## RECONSTRUCTION OF mtDNA AND NRY GENEALOGIES

Although the reconstruction of genealogies of mtDNA and NRY are broadly similar, there are some notable differences, probably owing to social differences in migration customs. For example, patrilocal marriage has historically been more common than matrilocal, which can explain differences in mtDNA and Y chromosome data in a number of populations (Cavalli-Sforza and Feldman, 2003). Demographic differences between the sexes, such as greater male than female mortality, the greater variance in reproductive success of males than females and possibly the greater frequency of polygyny than polyandry, may explain the discrepancy between the NRY and mtDNA dates (Cavalli-Sforza and Feldman, 2003).

Variation in mtDNA has been assessed mainly by using two different approaches: high-resolution analysis of the whole genome using restriction enzymes (restriction fragment length polymorphisms, RFLPs) or sequencing of the hypervariable control region (Santos et al., 2004). Used independently, both methods have shown that mtDNA evolution produces groups of phylogenetically related haplotypes that have been designated either by haplogroups or by clusters, respectively (Santos et al., 2004). More comprehensive studies have included the analysis of RFLPs and control region sequences for the same samples (Santos et al., 2004). As expected, and because of the lack of recombination in mtDNA, the groups of mtDNA types defined by each method were correlated. These studies have also shown that the two types of analysis are complementary; and their combined use has helped to clarify the phylogenetic relationship between mtDNA lineages (Richards et al., 1998; Macaulay et al., 1999). This has led to the unification of the nomenclature by adopting the system initiated by Torroni and coworkers (1992), which is based on RFLPs. (Santos et al., 2004)

Binary polymorphisms on the Y-chromosome are highly informative anthropological markers for reconstructing the prehistory of men (Underhill et al., 2000). The Y-Chromosome Consortium (YCC) has inferred a detailed tree of global Y-chromosome diversity from over 250 polymorphisms (Y Chromosome Consortium, 2002; Jobling and Tyler-Smith, 2003), and currently recognizes 18 primary branches called haplogroups or paragroups (labelled A to R) (Cox, 2006). One or more unique polymorphisms characterize each major monophyletic clade, whose distributions are often geographically restricted (Cox, 2006). Most Y chromosome studies first classify male samples to a major branch of the Y-chromosome tree, followed by more detailed classification via screening of additional single nucleotide polymorphisms (SNPs) and short tandem repeat (STR) sequences (Cox, 2006). A hierarchical

strategy like that suggested by Paracchini and co-workers (2002) and considered in forensics by Brion (2004), and in anthropological studies by Cox (2006) must be followed. This approach maximizes cost- and time effectiveness with information deriving from the lineage pattern of the sample studied. Since this strategy means that several loci must be examined, in order to identify the most suitable ones, multiplex PCR is preferable, starting from the minimal amounts of DNA which can usually be recovered for forensic analysis (Onofri et al., 2006)

## CASE STUDY: THE CAPE MUSLIM COMMUNITY OF SOUTH AFRICA

The Cape muslim community is one of the small communities living in South Africa. Although small, this community reflects the same diversity that characterizes the rest of the South African population. The history of this community is a key to understand many important chapters in the history of South Africa (Da Costa, 1990, 1992, 1994; Shell, 1994, 2000). The very existence of this community is linked to the slavery trade, migration, colonialism, ancient trade routes in Africa, inter-community marriages, and the spread of Christianity and Islam in the region.

The earliest Cape muslims were part of involuntary migration of slaves, political prisoners and criminals from Africa and Asia that lasted from 1652 to 1834 (Da Costa, 1994). Included in these people were the “Mardykers”, the Malay servants of Dutch officials who were in their way back to the Netherlands from the East. Many of these servants opted to remain at the Cape (Da Costa, 1994). The main group of African immigrants came from East Africa (Mozambique, the East African coast of the north, and Zanzibar), Madagascar, and West Africa; while the Asian immigrants came from India, Ceylon and the East Indies (Da Costa, 1994). In addition to migrations, this community has grown by combined and continuous multiplex processes of intermarriage, natural increase, conversion and blending with other communities (Shell, 1993; 2000).

South African muslims afford the historian the prospect of studying a well-documented, highly urbanized set of minority communities in a plural, modernizing society over three centuries (Shell, 2000). Indeed, researchers working on the history of this community have taken advantage of the availability of the huge amount of archival material covering the past three hundred years of the South African history. However, research on this community and other communities should also take advantage of the availability of modern molecular genetics tools. An integrated approach that uses both historical records and genetics can be of great benefit in the analysis of the social history of the Cape muslim community living in the Western Cape. The coming to existence and growth of the various small muslim communities that make up the larger muslim population in the Western Cape can be studied. Muslim communities who migrated from Cape Town to other cities of South Africa such as Port Elizabeth, East London, Kimberley, Mafeking, Johannesburg and Durban, can also be stu-died. The study can indeed complement previous studies on the history of the muslim population, and provide answers regarding its interactions with the other commu-nities living in the country. The contribution of these latter communities to the gene pool of the muslim population through intermarriages, conversion and blending can be analyzed.

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