

A molecular approach to the study of the ancient populations of southern Italy

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Abstract – The peopling of southern Italy must be traced back to the first expansion of the early forms of our species because of the geographical location of this area in the middle of the Mediterranean basin. Since then, the area was the site of a complex network of migration of different peoples, mainly from Phoenicia, Greece, Arabia, the Balkans, the Middle and Near East and north Africa, resulting in a heterogeneous pattern of both cultural and genetic interactions. In order to test the extent of the genetic impact of these various populations upon the peoples of southern Italy, a mitochondrial-DNA (mtDNA)-based research program was undertaken. This would help trace the geographic origin and past history of these populations.

Previous detailed and extensive investigations on the distribution of traditional marker allele frequencies in the Sicilian and Apulian extant populations, could only clearly detect the presence of the more recent gene flow from north Africa and the Middle East, superimposed on a predominant Greek genetic substratum. No traces of the ancient groups who peopled Italy in pre-Roman times were detected. Other authors still claim that it is possible to determine the existence of such ancient traces in southern Italy, stressing the difficulties connected to the reconstruction of human evolution based uniquely on the present structure of the genes.

Until recently, population genetic studies have heavily relied upon indirect evidence from the genetics of extant populations as well as information gained from cultural, historical and anthropological sources. The possibility to recover DNA from ancient soft tissue, bone and teeth, and the development of sensitive molecular techniques, such as PCR, have enabled us to directly compare genetic diversity between extinct populations and their extant counterparts. This has also provided a temporal dimension to the study of molecular evolution. These comparisons will generate independent data to address archaeological issues concerning migration, population replacement, lineage extinction, inbreeding, and the genetic relationships of ancient human populations.

DNA from bone and tooth samples obtained from the necropolises of Riferiscolaro (6th century BC) and Ticchiara (Bronze age), and DNA of extant populations from various Mediterranean areas, were extracted and analysed for mtDNA polymorphisms (region V, D-loop I and II). Moreover, in order to identify the remains of the family of the Prince Branciforte (a Sicilian maecenas and benefactor who lived between 16th and 17th centuries), molecular sex identification and mitochondrial DNA (mtDNA) analyses were used. a-DNA were extracted from the ribs of two adult and three young individuals.

NJ and ML phylogenetic reconstructions, performed to verify the reliability of the identified ancient lineages, confirmed the clustering of the Mediterranean mtDNAs into distinct lineage groups, each defined by a set of associated polymorphisms. The joint analysis of D-loop I and II allowed the identification of at least three distinct lineages within the previously identified aplogroup 1 (or H).

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