GENOMICS OF HUMAN AND ANIMAL POPULATIONS

Dr Olivieri is a member of the research group "***Genetics and Genomics of Human and Animal Populations***". The main general objective of the group is the reconstruction, based on genetic and genomic data, of the evolutionary history and demographic events that have involved present and past human populations (at both micro and macro-geographic levels) and some animals (especially domestic animals and those living in close contact with our species). Genetic findings can be easily employed also in multidisciplinary studies involving apparently far away scientific and cultural contexts, from forensics to history, archaeology, linguistics, anthropology, education and public health.

The genetic systems under study are the autosomes as well as the uniparentally transmitted mitochondrial DNA (mtDNA) and male-specific portion of the Y chromosome (MSY). The latter two are not reshuffled by recombination and thus constitute a molecular archive of the history and migration of females and males, respectively, who transmitted them to subsequent generations. Some of the research activities has also implications in biomedicine. Indeed the mitogenome plays an important role in bioenergetics, thus its sequence variation is involved in diseases, aging, athletic performance as well as environmental adaptation.

The current research activity of Dr. Olivieri is mainly focused on the following topic(s):

1) Origin of populations from Europe and the Mediterranean area

The demographic and genetic history of Europe and surrounding regions is extremely complex. The purpose of this research is to investigate, through the analysis of modern and ancient DNA genomes, what aspects of the variability of European populations can be traced back to the first colonization of the continent by modern humans, to post - glacial expansions, the Neolithic diffusion and (or) more recent events of gene flow. Over the years our studies on this general topic have been very productive and have used as the main tool of investigation the two non-recombinant genetic systems (mtDNA and MSY), which we are now integrating with genome-wide analyses.

2) The genetic origin of Italian populations

Despite its small geographic area, the Italian peninsula, for geographical, climatic and historical reasons, has occupied for several millennia a central position in the flow of genetic exchange and commercial trades among ancient populations. The objective of this research line is to reconstruct the prehistorical and historical genetics of the early Italy at a very high level of genetic resolution in a very large number of subjects (covering all Italian regions). To accomplish this task, a genome-wide scan on rare and common SNPs as well as analyses of both uniparental genetic systems (MSY and mtDNA) are going to be performed.

3) The peopling of Sardinia

Sardinian people are a “genetic outlier” in the European contest. Archaeological data point to a first evidence of modern human presence on the island dating back to about 13,000 years ago (Upper Palaeolithic). Distinctive genetic traits of Sardinians have been retrieved with all genetic systems, due to a combination of founder effects, geographic isolation, and selection. Recent paleo-genomic data based on nuclear genome markers have revealed that, among present-day populations, Sardinians retain the highest levels of genetic similarity with Early Neolithic farmers across Europe. However, this scenario appears in contrast with data provided by uniparentally-transmitted genetic systems. We are currently investigating on this topic, also from an ancient DNA perspective.

4) Identification of new pathological mtDNA mutations and the roles of mitochondrial backgrounds (haplogroups) in disease expression and environmental adaptation

The mitochondrial ATP production by the oxidative phosphorylation is essential for the maintenance of normal functions of organs and tissues. MtDNA mutations, by interfering with ATP synthesis, can cause serious maternally-transmitted diseases. The search for new disease-causing mtDNA mutations is carried out mainly on Leber's Optic Neuropathy (LHON). In recent years, an important role of the "neutral" sequence variation of mtDNA has also been postulated for many complex diseases and other phenotypes (aging, athletic performance). To evaluate also these aspects, we are sequencing entire mitogenomes from numerous human populations and building up a database that includes representatives of each mtDNA haplogroup and sub-haplogroup present in our species.

5) Origin and spread of the Asian tiger mosquito (Aedes albopictus)

Over the last 40 years, the tiger mosquito *Aedes albopictus*, indigenous to East Asia, has colonized every continent except Antarctica. Its spread has become a growing public health concern, being a competent vector for many arboviruses including those causing West Nile fever, yellow fever, Dengue fever, Saint Louis encephalitis and the recently emerged Zika fever.
The present study involves the definition and analysis of mitogenome sequence variation in samples from numerous African, European and American populations, in order to identify the ancestral Asian source(s) of *Ae. albopictus* adventive populations and their possible selective advantages as well as migration routes and expansion times.