

Prof. **Ornella Semino** 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 Prof. Semino is mainly focused on the following topics:

### ***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) The peopling of the Americas: a genetic perspective***

The first peopling of the New World is a hot topic in genetics and anthropology. Archaeology, linguistics and genetics agree on the Asian origin of Native Americans. However, the arrival times of the first settlers, the number of migrations / expansions involved in the process and in the subsequent colonization of Central and South America, as well as the genetic consequences of the flourishing of the great Mesoamerican and Andean civilizations are still extensively debated. To clarify these issues, sequence analyses of the two non-recombinant genetic systems (MSY and mtDNA), both at the micro- and macro-geographic levels, are carried

out in several modern populations and some ancient samples. In some contexts, these studies are integrated by genome-wide analyses at the nuclear level.