

Study of cattle phylogeny by means of mitochondrial DNA (mtDNA) and Y-chromosome variability

Bovine mtDNA variability

The domestication of *Bos primigenius* – happened ~10,000 years ago – was one of the major innovations in the Neolithic “revolution” and had significant effects on the cultural and socio-economical life of Old World populations that adopted farming. We have been studying the process of domestication since few years by analysing a large dataset (~2,000) of bovines belonging to European, Near-East and African breeds for variability at the mtDNA control region and extending the analysis to >100 complete mtDNA genome sequences of particular interest. In doing so we found modern cattle harboring mitochondrial genomes that derive from the extant European *Bos primigenius*, and belong to distinct haplogroups (P and Q) that show a somewhat different geographical distribution. In addition we later identified in modern Italian cattle a rare haplogroup (R) that was previously unknown and predates both P and Q haplogroups. This led us to hypothesise that in historical times an additional and independent domestication event might have occurred in Italy.

In summary our research is aimed at enriching the phylogeny of bovine populations by using the most detailed information available, i.e. the complete sequence of mtDNA, and unveil haplogroups that represent the legacy of the extant *Bos primigenius* in modern populations, with special care to autochthonous breeds that might have maintained a higher level of genetic diversity.

Isolation of novel biallelic markers for the analysis of bovine Y-chromosome variation

The goal of the project is to enrich the number of available biallelic markers employed to study the variation of bovine Y-chromosome. Our approach is straightforward: using the available collection of bovine Y-specific BAC clones, and starting with currently used Y markers we built a contig of clones that we mine for additional STS. The potential STS are first selected based on the absence of repetitive DNA, appropriate size for an easy PCR, the Y-specificity and are later scored for the presence of single nucleotide polymorphisms in a panel of bovines representing a range of Italian and European breeds. DHPLC is used for the initial scoring of polymorphisms that are later confirmed by sequencing. Our preliminary results showed 12 new SNPs some of which allow the separation of haplogroup Y2 in two sub-haplogroups, Y2A and Y2B. We also identified a novel STS that thanks to a 81 bp indel can be used to quickly assign Y chromosomes to the Y1 or Y2 haplogroups with a simple PCR in place of sequencing for several STS as in the current typing protocol.