Mitochondrial DNA as a Marker for Genetic Diversity and Evolution

Othman El-Mahdy Othman

The formulation of the modern breed concept during mid-1800s has caused remarkable changes in the livestock sector: large-scale production expanded [1] and its application to breeding and husbandry practices led to the formation of well-defined breeds, exposed to intense anthropogenic selection. As a consequence, farmers progressively substituted the less productive, locally adapted, native breeds with highly productive cosmopolitan breeds and progressively abandoned marginal areas [2].

Therefore, a significant number of cattle, sheep, and goat breeds disappeared and many are presently endangered. Twenty percent of the breeds world-wide are classified as being critically endangered, critically-maintained, endangered, or endangered-maintained [3]. It is likely that a high number of breeds are being, and will be lost in the near future, before their characteristics can be studied and their potential evaluated. In these conditions it is more strategically important than ever to preserve as much the farm animal diversity as possible, to ensure a prompt and proper response to the needs of future generation.

Mitochondrial DNA sequencing has been used to explain the origins of many modern domestic livestock species. The existence of multiple mtDNA lineages and their mixing within breeds could be due to multiple domestication events or to introgression between domestic and wild species. The studies on the structure and function of mtDNA become highlights in the research area of molecular evolution, classification, population genetic analysis, relative identification and quantitative traits loci [4-7].

Mitochondrial DNA contains 37 genes, all of which are essential for normal mitochondrial function. Thirteen of these genes provide instructions for making enzymes involved in oxidative phosphorylation. The remaining genes provide instructions for making molecules called transfer RNAs (tRNAs) and ribosomal RNAs (rRNAs), which are chemical cousins of DNA. These types of RNA help assemble protein building blocks (amino acids) into functioning proteins.

In most animal species, mitochondria appear to be primarily inherited through the maternal lineage. Typically, a sperm carries mitochondria in its tail as an energy source for its long journey to the egg. When the sperm attaches to the egg during fertilization, the tail falls off. Consequently, the new organism has only the mitochondria which came from the egg of its mother. Therefore, unlike nuclear DNA, mitochondrial DNA doesn’t get shuffled every generation, so it is presumed to change at a slower rate, which is useful for the study of organism evolution. Mitochondrial sequencing has been used to illustrate the genetic diversity and molecular evolution in livestock sector, for example goat and sheep breeds.

Mitochondrial DNA studies on domestic goats identified at least four major mtDNA lineages. Lineage A is the most diverse and widely distributed across all continents. Lineage B is confined to eastern and southern Asia, including Mongolia, Laos, Malaysia, Pakistan and India. Lineage C is present in low frequencies in Mongolia, Switzerland, Slovenia, Pakistan and India. Finally, lineage D is rare and only observed in Pakistani and Indian local goats. The time since divergence among these four lineages (more than 200,000 years ago) far predated the time of domestication around 10,000 years ago [8-11].

Both archaeozoological and genetic evidences indicated that the domestication of wild sheep occurred 8000-9000 years ago. Most of the information about history and domestication of this species have been gathered using mtDNA. Mitochondrial DNA analysis in sheep identified an increasing number of maternal lineages: two [12-14], three [4,15], and then five [7]. The main haplogroups A and B are both found in Asia, while B dominates in Europe. Haplogroup C has been found in Portugal, Turkey, the Caucasus, and China [16]. Whereas haplogroup D, present in Rumanian Karachai and Caucasian animals, is possibly related to haplogroup A.

References


Author Affiliations

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1. Professor, Cell Biology Department, Genetic Engineering and Biotechnology Division, National Research Center, Egypt