Mitochondrial DNA (mtDNA) – What Is It and What Does It Tell Us?

The Basics – Nuclear DNA vs. mtDNA

Deoxyribonucleic acid (DNA) is the hereditary material passed from generation to generation, which provides the blueprint for what an individual will look like and how its body systems will function. DNA is made up of chemical bases; hundreds or thousands of base pairs form a gene sequence, which is the basic unit of heredity. Genes provide the instructions (also known as a "code") for the production of a variety of proteins; the proteins, in turn, determine how a cell operates and what physical traits will develop.

The most commonly discussed type of DNA is the nuclear DNA (named for its location in the cell's nucleus), which is contributed by both parents; the sire and dam each contribute 32 chromosomes that contain this type of DNA. A horse receives half of its nuclear DNA from the sire and the other half from the dam. Each half represents a shuffled recombination of DNA that has been passed down from ancestors through the generations. Because of the DNA recombination which occurs each generation, it is difficult to use nuclear DNA to study lines of descent more distant than direct parentage.

A second type of DNA is the mitochondrial DNA (mtDNA), which is contributed by the dam. Unlike the nuclear DNA, which is contained in the cell's nucleus, mtDNA is found in the cell's mitochondria. Mitochondria are structures in the cell's cytoplasm that convert energy from food into a usable energy form for the cell. Because mtDNA is contributed through the dam line only, it is a powerful tool for tracking ancestry through the tail female line of a pedigree.

In addition to being located in different parts of the cell, nuclear DNA and mtDNA also have different structures. Nuclear DNA molecules are formed as a spiral staircase with each stair composed of DNA bases: adenine (A) pairs with thymine (T) and cytosine (C) pairs with guanine (G). Horses have approximately 2.7 billion base pairs of nuclear DNA of which only 2-3% makes up the approximately 20,000 genes contained in the equine genome.

On the other hand, mtDNA is a circular chromosome with about 17,000 base pairs, most of which code for replication activity. In addition, mtDNA does not undergo recombination, so there is no shuffling of DNA as it is passed from generation to generation. Due to a lack of recombination, the mtDNA code is varied only through occasional mutations. When a mutation occurs and is passed along, it becomes a marker of descent. Since mtDNA is passed only from the dam and is inherited completely independent of the nuclear DNA, it allows for the study of tail female lineage back to a common ancestral point.

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Table: 1 Comparison of nuclear DNA and mtDNA

DNA Type	*Location*	*Number of Base Pairs*	*Condributed By*	*Structure*	*Recombination*
Nuclear	On 64 chromosomes (32 pairs) in the cell's nucleus	~2.7 billion	Sire and Dam (each contributes 32 chromosomes)	Linear Double Helix	Yes
Mitochondrial (mtDNA)	In mitochondria located in the cell's cytoplasm	~17,000	Dam Only	Circular Double Helix	No

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