EDITORIAL

Mitochondrial DNA in human identification: A Review

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Human genetic identification for forensic purposes is achieved through the definition of genetic profiles. A genetic profile or the genetic fingerprint of an individual is the phenotypic description of a set of genomic loci that are specific to that individual[2]. In accordance with international recommendations, particularly with recommendations of the European DNA Profiling Group (EDNAP), currently, only genetic profiles obtained from autosomal short tandem repeats (STR) should be used for genetic fingerprinting[1]. Mitochondria are cellular organelles that contain an extra chromosomal genome, which is both different and separate from the nuclear genome. Mitochondrial DNA contains 37 genes, all of which are essential for normal mitochondrial function[2,4]. Thirteen of these genes provide instructions for making enzymes involved in oxidative phosphorylation. Oxidative phosphorylation is a process that uses oxygen and simple sugars to create adenosine triphosphate (ATP), the cell's main energy source. The remaining genes provide instructions for making molecules called transfer RNA (tRNA) and ribosomal RNA (rRNA), which are chemical cousins of DNA. These types of RNA help assemble protein building blocks (amino acids) into functioning proteins[2].

Human mitochondrial DNA (mtDNA) accounts for a small portion of his total DNA. It contains just 37 of the 20,000 to 25,000 protein-coding genes in human body. But it is notably distinct from DNA in the nucleus[6]. Unlike nuclear DNA, which comes from both parents, mitochondrial DNA comes only from the mother[1]. There is a question that why or how fathers' mitochondrial DNA gets wiped from cells? An international team of scientists recently studied mitochondria in the sperm of different sets of human beings and concluded that sperm mitochondria become inert by some mutational aspects during the process of fertilization[3].

Human mitochondria and mtDNA are maternally inherited. This is probably because the mid-piece of the sperm, which is the only part containing mitochondria, does not penetrate the ovum during fertilization[4]. There is some evidence for a small paternal contribution, but to date this does not seem to have any role in disease pathogenesis. Because mtDNA is maternally inherited, mtDNA mutations have the potential to be inherited in a matrilineal fashion. In practice, most mtDNA point mutations are maternally inherited[6]. In contrast, large-scale rearrangements of mtDNA, such as large deletions, are not usually maternally inherited. There is also a small group of recently recognized mtDNA point mutations that are not maternally inherited. The precise explanation why certain mutations are inherited maternally and others are not is unclear. For maternally inherited mutations, a mother harboring the mtDNA mutation will transmit the mutant mtDNA to all her offspring. However, this does not imply that all offspring will develop disease. A number of factors seem to be important in determining the penetrance of an mtDNA mutation in a given individual[3]. These include the amount of the mutant mtDNA and its tissue distribution, the mtDNA haplotype, the mtDNA copy number, and the nuclear background[2].

The mtDNA is maternally inherited. Even though a zygote receives both maternal and paternal mtDNA at fertilization, the paternal mtDNA is specially targeted for elimination and removed from the cytoplasm of the zygote during very early embryogenesis[5]. The amount of mitochondria present in a metaphase II oocyte correspond to only a small portion of the maternal mtDNA pool due to a genetic bottleneck that takes place in the course of oogenesis. In the primordial germ cells there is a great population of mtDNA that represents the maternal mtDNA pool[6]. This is the starting point of the genetic bottleneck. At some stage in development of the germline, the maternal mitochondrial pool is subsampled to a quite small number resulting in only a small percentage of mtDNA being represent in the mature egg. In this way, the variety of mtDNA is restricted in each oocyte, and homoplasmy is promoted[4]. Therefore, within a cohort of oocytes there are differences for the reason that the maternal mtDNA is arbitrarily segregated; in fact, in mtDNA diseases the level of mutant mtDNA differs in oocytes from the same patient[2]. Subsequent to the bottleneck, during oocyte maturation, there is an augment in mitochondrial content and mtDNA copy number[6].

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