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Role of Mitochondrial DNA in Human Evolution and Migration

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Abstract:

Many years ago, when the population was being increased day by day, then the people started to migrate from their places and lived to another places. Before some decades, many scientists and researchers wanted to know how the evolution took place and where the people migrated from their origin place. For this they studied the histories and different methods. After many researches and studies, the molecular biology was used for the purpose to know the human evolution and their migration. In the molecular biology, Mitochondrial DNA is considered as the best method as it has many unique features such as maternal inheritance, lack of recombination, several copies of mtDNA in mitochondria, and high mutation rate as compared to the nuclear DNA. And mtDNA is resistant to degrade and more stable in unfavorable conditions. This paper discuss about the review of many author and researchers about the human evolution and their migration through the analysis of Mitochondrial DNA.

Keywords: Mitochondrial DNA, Evolution, Migration, Molecular Biology

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Introduction

It is believed that the homo genus are originated in Africa. The Homo erectus group was migrated from Africa to another place such as Europe and Asia within the one or two million years. Homo erectus changed their appearance and character because of the different reasons. Homo erectus is called by the name as archaic by anthropologist. They involved Neandertals who were living in Middle East and Europe form 150, 000 to 28, 000 years.

The process of evolution is very difficult and has the long history as it started from the first appearance of the human and being continued today. Before some decades, there was only one way of studying the evolution and migration of human that is uncovered skeletal remains. In last two decades, the anthropologist gained various new tools, in which molecular biology is one of them. In the molecular biology, proteins like biomolecules' history and evolution were analyzed but they were not enough to give the precise result regarding evolution. After that the researchers insight for the DNA. The main key of the human evolution is the changes in the gene pool and through the study of genetic origin of populations, we are able to understand the population relationship as well as the alteration in the gene with the space and time. Actually, DNA is a biomolecule which reveals the history record of the evolution of life on earth. Most of the DNA is located in nucleus of the cell, small quantity of the DNA is present also in another cell organelle that is mitochondria.

Mitochondria plays a significant role in cell energetics, apoptosis, ageing and other pathological conditions. It also contains DNA, is called as Mitochondrial DNA (mtDNA). Mitochondrial DNA is one of those tools which is used to determine the origin of population because it has the high evolutionary importance.

For the study of human evolution through mitochondrial DNA, the best ways to collect the samples are museum specimens, fossil remains and archaeological finds as they can provide the evidence related to their origin and migration processes by comparing it with the modern mtDNA. In the collection of ancient mitochondrial DNA, the researchers face the problems related to the contamination and authenticity of DNA. In spite of having these issues, there is a settled criteria and platforms for collecting authentic ancient DNA **(Nesheva, 2014)**.

The first successful extraction and sequencing of the ancient mtDNA from the dried muscle was done by Higuchi et al. This dried muscle was 150 old museum specimen of the quagga, a species of zebra. In spite of many studies, the doubt was remain about the achievement of investigations regarding DNA studies. After that the development of anthropogenetics and paleogenetics and their increased role gave the new insight in the field of evolutionary science.

In the analyses of paleogenetic, the extracted mtDNA form the fossil and skeletal remains found form the archaeological site are sequenced through their hypervariable fragments by scientists. After the analyses, the data is compared with the literature data and then classified mtDNAs into maternal lines as per the polymorphism sequences.

Mitochondrial DNA

It is present in the form of the circular double stranded molecule having 16,569 base pairs. These base pairs are coded for 13 subunits of oxidative phosphorylation system, 22 transfer RNAs (tRNAs) and 2 ribosomal RNAs (rRNAs) through which translation of proteins are taken palce in the organelle. Each cell contains hundred to thousand copies of mitochondrial DNA outside the nucleus but in the energy generating cell organelles, mitochondria. Mitochondrial DNA is considered as a vital source of study in various field i.e., population genetics, evolutionary anthropology, genetic genealogy, medical genetics and forensic science. There are some unique features of mtDNA that make it amenable to study the evolution. In these features high copy number, lack of recombination, maternal inheritance and high mutation rate are included which are not seen in nuclear DNA.

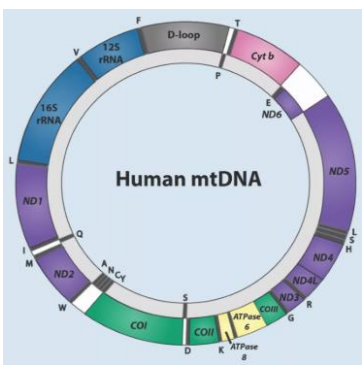

Figure: Mitochondrial DNA

High Copy Number

Somatic cell has only two copies of DNA segment or nuclear gene while mitochondria contains hundred to thousand copies of mtDNA. This property including the extra nuclear, cytoplasmic location of mtDNA, makes possible to extract mtDNA easily from the ancient DNA and their analysis along with other forensic DNA application. The mtDNA is exist in different types within an individual, is known as heteroplasmy. There are two types of heteroplasmy – length heteroplasmy and sequence heteroplasmy.

Length heteroplasmy exist around the homoploymetric C-stretch in HVI at the location between 16184 and 16193 and HVII at the location between 303 and 31. While Sequence heteroplasmy is analyzed by the occurrence of two nucleotides in sequence electropherogram **(Samehsalari and Reddy, 2018)**.

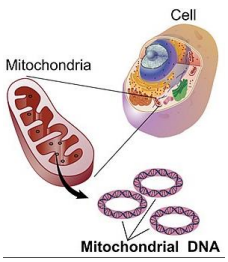

Figure: Multiple copies of mtDNA

Maternal Inheritance

In the case of mtDNA, the maternal inheritance is considered as a constant philosophy of the field and also known as uniparental mode of DNA transfer. During the reproduction process, the mitochondrial DNA is only contributed by the egg for developing embryo. Few mitochondria are contained by the sperm head, but these are destroyed after fertilization by ubiquitin and allow only maternal inheritance. The advantage of this uniparental mtDNA is useful in tracing the lineages back in the time.

Lack of Recombination

Molecular anthropology showed another theory that the mtDNA is not undergo recombination. But this theory was failed after receiving four papers regarding the recombination in mtDNA. In these four papers, three paper were based on the phylogenetic and statistical evaluation of mtDNA sequences and the fourth study showed direct evidence of recombination in Melanesia. Though these studies had been failed because of the faulty data and questionable methods of statistics, and alignment error. Now recently, there was another case where the recombination in human mtDNA was seen in known human with both paternal and maternal mtDNA. In this case, only in 0.7% of total mtDNA present in the muscle tissue of patient showed the recombination between the maternal and paternal mtDNA.

Mutation Rate

Mitochondrial DNA has the high rate of mutation compare to the nuclear genes. In the mtDNA, there are two hypervariable regions of the noncoding control region that are HVR I and HVR II which show high rate of mutation. The difference between phylogenetic and pedigree based approximations of mtDNA mutations rate can be understand through the observation as in the control region, the rate of mutation is in heterogeneous form which have some mutational hot spots and mutate four to five times faster than the average site **(Pakendorf and Stoneking, 2005)**.

The mutation in the mitochondrial DNA does not have the repair mechanisms due to which it slow down the mutation rate of nuclear genome. Hence, it creates a high level of sequence variation among the individuals, accumulated in short duration as compare to the ancestral mtDNA molecule **(Witas and Zawicki, 2004)**.

Neutral mtDNA Variants

In the geographical separated populations, the mtDNA sequence is varied with high degree. Firstly, it is detected by the restriction fragment length polymorphisms (RFLPs) in which most of this variation are neutral including nucleotide replacements in the third codon locations as well as in noncoding regions like control region. The sequence variation of mtDNA was used to correlate with the ethnic and geographic origin of an individuals through the analysis of Hpa I RFLPs extracted from the samples taken from the Aisa, Europe and Africa **(Wallance, 1994)**.

Review of Literatures

Samehsalari and Reddy (2018), proposed about mitochondrial DNA/genome, they talked about the forensic value of mtDNA. Mitochondrial DNA is present in number of copies in cell organelle, mitochondria due to which it does not degrade and

stay stable in worse conditions as compared to the nuclear DNA. They also explained about the control region or d-loop which allow the individuals to separate with others. The mtDNA can be extracted from the skeletal remains and fossils and play a significant role in forensic anthropology. As it does not have the issues of contamination because of the present in large quantity (numbers of copies).

Rishishwar and Jordan (2017) concluded on the basis of their analysis done on the human genetic variation that divergent human populations' nuclear and mitochondrial genomes can coexist in healthy individuals. This statement indicates that these mismatched nuclear DNA and mitochondrial DNA mixtures are not damaging and also not removed by purifying selection. Among the human population, the levels of nDNA and mtDNA sequence deviation are correlated, consistent with their co-evolution. While there are many cases where nuclear and mitochondrial genomes from the divergent populations are co-occurred within individual humans.

According to Pakendorf and Stoneking (2005), mitochondrial DNA has some unique characteristics like high copy number of mtDNA, lack of recombination, high mutation rate and maternal inheritance which make it a choice of study for the analysis of human population history and their evolution and migration. In their paper, they reviewed these characteristics and concluded that the evolution and migration of human populations can be studied through the mitochondrial DNA. Mitochondrial DNA studies are added with the analyses of y- chromosomes and nuclear DNA variation. They also told about the some serious issues which should be addressed regarding nuclear inserts, database excellence and possible effect of selection on mtDNA deviation. They also discussed about the application of mtDNA in different fields as ancient DNA, human genetic variation and the impact of social culture on them, certain forensic DNA application and tracing personal genetic history.

Witas and Zawicki (2004), stated that the techniques of molecular biology were used to broaden the evolution and anthropology. In their paper, they concluded that the migration compare to the male affect the structure of nuclear genes. This type of situation comes when the offspring migrate with their mothers during pregnancy and lactation. According to them, people with new variants may take some time in settling after the creation of diversity. They also suggested that the dating of human remains, study of higher numbers of heritably identified samples and emerging the new molecular methodologies can bright the hiding history.

Nesheva (2019), For describing a prehistoric and historic events, anthropologists and geneticists created routes of human migration as well as the time scale. They compared the ancient and modern data for knowing human history since 200,000 years ago in east Africa. According to them, if we combined the present and future data, we would be able to make a map of ancient people and relationships among them. The method for generating the complete and clear picture of the history of human evaluation is very complicated due to which it needs further analyses in those places where DNA has not been graphed.

Conclusion

After studying the reviews of other researchers, this paper concluded that the mitochondrial DNA is an important source for determining the human evolution and their migration to different places. Actually, the mtDNA has many vital features which make it different form the nuclear DNA. These vital features are maternal inheritance, lack of recombination, number of copies of mtDNA and last one is high mutation rate. Some researchers raised a questioned about authenticity of the mtDNA during the analysis of skeletal remains and fossils. But despite of this it is used for the human evaluation analysis. For the comparison of ancient mtDNA and modern DNA, the old sample are collected from the museum. But the process of analysis of mtDNA from the human skeletal, fossil and other source are very difficult task for the analyst. So, there is need to search the other methods for the determination of human evolution and migration.

References:

"Molecular Anthropology - Mitochondrial Dna and The Origin Of Modern Humans." *Chromosome, Genetic, Populations, and Recent - JRank Articles*, medicine.jrank.org/pages/2546/Molecular-Anthropology-Mitochondrial-DNA-Origin-Modern-Humans.html.

Cann, Rebecca L., et al. "Mitochondrial DNA and Human Evolution." *Nature*, vol. 325, no. 6099, 1987, pp. 31–36., doi:10.1038/325031a0.

D.v., Nesheva. "Aspects of Ancient Mitochondrial DNA Analysis in Different Populations for Understanding Human Evolution." *Balkan Journal of Medical Genetics*, vol. 17, no. 1, 2014, pp. 5–14., doi:10.2478/bjmg-2014-0019.

Pakendorf, Brigitte, and Mark Stoneking. "Mitochondrial Dna and Human Evolution." *Annual Review of Genomics and Human Genetics*, vol. 6, no. 1, 2005, pp. 165–183., doi:10.1146/annurev.genom.6.080604.162249.

Rishishwar, Lavanya, and I. King Jordan. "Implications of Human Evolution and Admixture for Mitochondrial Replacement Therapy." *BMC Genomics*, vol. 18, no. 1, 2017, doi:10.1186/s12864-017-3539-3.

Samehsalari, Somayyeh, and K. Rajasekhara Reddy. "Application Control Region of Human Mitochondrial DNA in Forensic Anthropology." *International Journal of Modern Anthropology*, vol. 2, no. 11, 2018, p. 233., doi:10.4314/ijma.v2i11.11.

Wallace, D. C. "Mitochondrial DNA Sequence Variation in Human Evolution and Disease." *Proceedings of the National Academy of Sciences*, vol. 91, no. 19, 1994, pp. 8739–8746., doi:10.1073/pnas.91.19.8739.

Witas, Henryk W., and Przemyslaw Zawicki. "Mitochondrial DNA and Human Evolution: A Review." *Przegląd Antropologiczny*, vol. 67, 2004, pp. 97–110.