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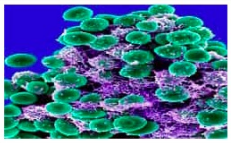
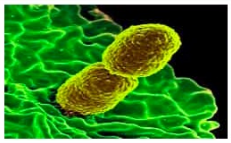

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Title : SIBLING ANALYSIS USING MITOCHONDRIAL DNA DISPLACEMENT LOOP (mtDNA D-LOOP) REGION IN THE IDENTIFICATION OF MADURESE POPULATION

Author : Ahmad Yudianto, Agung Sosiawan, Retno Palupi, Masniari Novita,

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SIBLING ANALYSIS USING MITOCHONDRIAL DNA DISPLACEMENT LOOP (mtDNA D-LOOP) REGION IN THE IDENTIFICATION OF MADURESE POPULATION

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Abstract— Analyzing the variations of nucleotide sequences in D-loop regions mtDNA can determine the identity of specific individuals or populations and maternal kinship. The Madurese population is an ethnic group that maintains its traditional customs in harmony with the religions they adhere to and endogamous marriage as their marriage pattern. This study applied PCR amplification and sequencing strategies on HVS-1 143 bp (nt 16268-16410) and HVS 2 126 bp (nt 34-159) of mtDNA D-loop regions. This study used buccal swab samples collected from 50 pure Madurese families consisting of a mother and two children. The homology analyses of female-female, male-female, and male-male siblings showed 11 variants or morphs in 126bp HVS-2 D-Loop mtDNA (nt 34-159). The highest variant were: female-female siblings (129G→C: 15%), male sibling (120C → A: 11.5%) - female sibling (120C: A: 11.5%), and male-male siblings (131T→C: 11.5). The homological analysis of female-female and male-female siblings showed 11 variants on 143bp HVS-1 mtDNA D-Loop (nt 16259-16410). The highest variants in female-female siblings were: 16387A→G, 16387A→C: 15%; male sibling-female sibling: 16393C→T, 16393C→A: 11.5%; while the homological analysis of male-male siblings showed 13 variants with the highest percentage: 16367A→G, 16367A→C: 11%.

Keywords: Mitochondrial DNA, variants, D-Loop, Siblings, Madurese Population, Human & Mortality

1. Introduction

Personal identity is an essential part of achieving proper justice. Five identification methods commonly used are fingerprints, dental, deoxyribonucleic acid (DNA), property, and medical identification methods. DNA analysis identifications involve somatic chromosomes, and a comparison originates from a mother or a father. Meanwhile, mitochondrial DNA analysis uses a maternal family member as the comparison.[1,2,3]. This unique heredity system has been applied in various aspects, such as determining kinship relationships, studies on human evolution and modern human migrations, forensics, and genetic disease identifications.[4]. The unavailability of information originating from parental or descendant sources that can be used in forensic DNA analysis has become one of the problems in forensic DNA analysis.

The principle of forensic DNA analysis of nucleic DNA is based on the process of allelic comparison with the alleles originating from the parents or family line sources (kinship analysis). In contrast, mitochondrial DNA analysis is based on the comparison process with one family member from the maternal line (maternal kinship). One of the applications of kinship is the usage of sibling analysis in forensic DNA identifications, both in nucleic DNA and mitochondrial DNA identifications of forensic cases, such as parentage testing (civil or criminal), disaster victim identification, missing person identification, and familial searching[5].

Madurese people are an ethnic group that still maintains most traditional customs in harmony with their religions. Marriages among Madurese people, especially those living in rural areas of Madura Island and its nearby islets, remain endogamous [6]. From the genetic point of view, endogamous marriage may increase the frequency of homozygote genotypes. Genetic homogeneity improves when endogamous marriages are repeated through generations to the point that homozygote alleles occur in a locus or all loci [5,7].

Therefore, it is of interest to the present study to determine the presence of novel morphs or variants in Madurese (different from the reference sequences, i.e., Cambridge or Anderson Reference Sequences) which later can be used as a reference in determining mtDNA genetic patterns of Madurese in a large scale.

2. Material and Methods

This study applied PCR amplification and sequencing strategies on HVS-1 143bp (nt 16268-16410) and HVS-2 126bp (nt 34-159) of mitochondrial DNA D-loop regions. Buccal swab samples were collected from 50 volunteers of pure Madurese families consisting of a mother and two children. The Ethical Committee of Universitas Airlangga Faculty of Dentistry issued permission to conduct this experiment. Every volunteer has read and agreed to the informed consent sheet before being included in this experiment. The inclusion criteria for the volunteers are based on the pedigree proving the three generations are pure Madurese population.

Sample Treatment

The preparation of DNA templates for PCR amplification: DNA templates originated from buccal swabs. The tip of buccal swabs is cut and put into conical tubes and mixed with Water Free. The tubes are centrifuged at 10,000 g for 10 minutes. The pellets were collected and mixed with 1 ml DNAZol and centrifuged at 10,000 g for 10 minutes at 4 °C temperature. 0.5 ml of absolute alcohol was added to the viscous supernatant and centrifuged at 4,000 g for 1-2 minutes at 4 °C temperature. The pellets were washed with ethanol 75% 0.8-1 ml. The shots containing DNA have diluted in 0.2-0.3 ml NaOH 8 Mm solution, vortexed properly, and stored at -20 °C temperature.

Polymerase Chain Reaction (PCR) Amplification

The materials for PCR amplification were: dNTP (ATP, CTP, TTP GTP), MgCl2, Taq Polymerase, Nuclease Free water, primer mtDNA 126 bp (nt 34-159. HVS-2) 5'- GGG AGC TCT CCA TGC ATT TGG TA-3' and 5'- AAA TAA TAG GAT GAG GCA GGA

ATC-3' and mtDNA 143bp (nt 16268-16410 HVS-1): 5' GGG AGC TCT CCA TGC ATT TGG TA 3'and 5' AAA TAA TAG GAT GAG GCA GGA ATC 3' [5].
The amplification program for 126bp mtDNA consisted of an initial denaturation phase at 95 °C-3 minutes and a 30-cycle phase comprised of denaturation at 94 °C-1 minutes, annealing at 56 °C-1 minutes, extension at 72 °C-1 minutes, and final extension at 72 °C-3 minutes. Meanwhile, the amplification of 143bp mtDNA consisted of an initial denaturation phase at 95 °C-4 minutes and a 35-cycle phase comprised of subsequent denaturation at 94 °C-1 minutes, annealing at 61 °C-1 minutes, extension at 71 °C-1 minutes, and elongation at 72 °C-5 minutes [8]. The results of PCR amplification were visualized using agarose gel electrophoresis (b/v).

Sequence Analysis

The sequencing process was carried out following ABI PRISM DNA Sequencing procedures. The dye terminator method's reaction consisted of several phases: DNA template preparation, PCR amplification process, DNA purification using Sephadex G-50 columns, acrylamide gel electrophoresis, electropherogram reading, and the analysis of sequencing results. An ABI PRISM DNA Sequencer device automatically reads the electrophoresis results [8,9]
Every nucleotide generates peaks with different colors on the electropherogram. A-nucleotides are green; G-nucleotides are black; C-nucleotides are green; T-nucleotides are red. The analysis of the finding was carried out using the DNA-STAR program. Analyses were also conducted on identity probabilities (frequency), homological studies on nucleotide sequencing among the samples, and comparison to CRS or Anderson sequences.

3. Results

The profiles of the samples were female-female siblings (40%), male-male siblings (35%), and male-female siblings (25%). The result of homological analysis on nucleotide sequences of the samples compared to the reanalysis of Cambridge Reference Sequences (rCRS)[9] on 126bp D-Loop HVS-2 was nt 34-159 (Tables 1, 2, and 3) and the result of homological studies of the sample against rCRS on 143bp D-Loop HVS-1 was nt 16268-16410. The sequences of the reference, 126bp D-Loop HVS-2 nt 34-159, and 143bp D-Loop HVS-1 (r-CRS) nt 16268-16410 are:
>NC_012920.1:34-159 Homo sapiens mitochondrion, complete genome
GGGAGCTCTCCATGCATTTGGTATTTTCGTCTGGGGGGTATGCACGCGATAGCAT
TGCGAGACGCTGGAGCCGGAGCACCCTATGTCGCAGTATCTGTCTTTGATTCCTG
CCTCATCCTATTATTT

>NC_012920.1:16268-16410 Homo sapiens mitochondrion, complete genome
CACTAGGATACCAACAAACCTACCCACCCTTAACAGTACATAGTACATAAAGCC
ATTTACCGTACATAGC
ACATTACAGTCAAATCCCTTCTCGTCCCCATGGATGACCCCCCTCAGATAGGGGT
CCCTTGACCACCATCCTC

Table 1. The variants of 126bp HVS-2 (nt 34-159) D-Loop mtDNA in the female-female siblings of Madurese Population (N: 20)

	Nucleotide (nt)										
	101	**107**	**109**	**110**	**118**	**129**	**130**	**131**	**138**	**143**	**155**
rCRS	**G**	**G**	**A**	**G**	**T**	**G**	**C**	**T**	**G**	**T**	**A**
Female	**G**	**G**	**T**	**G**	**A**	**G**	**C**	**T**	**T**	**T**	**G**
Female Siblings	**A**	**A**	**G**	**A**	**A**	**C**	**A**	**C**	**G**	**T**	**T**
%	**8%**	**7,5%**	**5%**	**10%**	**8%**	**15%**	**10%**	**5%**	**12%**	**8,5%**	**11%**

Table 2. The variants of 126bp HVS-2 (nt 34-159) D-Loop mtDNA in the male-female siblings of Madurese Population (N: 17)

	Nucleotide (nt)										
	54	**77**	**109**	**110**	**120**	**129**	**130**	**131**	**138**	**143**	**155**
rCRS	**G**	**A**	**A**	**G**	**C**	**G**	**C**	**T**	**G**	**T**	**A**
Male	**G**	**T**	**T**	**A**	**A**	**G**	**C**	**T**	**T**	**A**	**G**
Female Siblings	**A**	**T**	**A**	**A**	**A**	**C**	**A**	**C**	**G**	**T**	**T**
%	**3%**	**10,5%**	**10%**	**8%**	**11,5%**	**11%**	**13%**	**6%**	**10%**	**7%**	**10%**

Table 3. The variants of 126bp HVS-2 (nt 34-159) D-Loop mtDNA in the male-male siblings of Madurese Population (N: 13)

	Nucleotide (nt)										
	50	**67**	**105**	**110**	**125**	**130**	**131**	**135**	**140**	**143**	**155**
rCRS	**T**	**G**	**C**	**G**	**T**	**C**	**T**	**T**	**C**	**T**	**A**
Male	**T**	**T**	**T**	**A**	**A**	**C**	**T**	**T**	**T**	**A**	**G**
Male Siblings	**A**	**T**	**C**	**A**	**A**	**T**	**A**	**C**	**C**	**T**	**T**
%	**8%**	**10%**	**5,5%**	**8,5%**	**8%**	**11%**	**11,5%**	**8,5%**	**10,5%**	**8,5%**	**10%**

The results of homological analysis of female-female siblings, male-female siblings, and male-male siblings indicate 11 variants or morphs on 126bp HVS-2 D-Loop mtDNA (nt 34-159). The variants with the highest percentage are (129G→C: 15%) in female-female siblings (Table 1), (120C→A: 11.5%) in male-female siblings (Table 2), and (131T→C) in male-male siblings (Table 3).

Table 4. The variants of 126bp HVS-2 (nt 34-159) D-Loop mtDNA in the female-female siblings of Madurese Population (N: 20)

	Nucleotide (nt)										
	16277	16289	16299	16320	16367	16387	16390	16398	16400	16401	16409
rCRS	A	A	A	C	C	A	G	G	C	C	T
Female	G	G	T	G	A	G	C	T	T	T	G
Female Siblings	A	A	G	A	A	C	A	C	G	T	T
%	5%	7%	5%	13%	10%	15%	10%	5%	10%	9%	11%

Table 5. The variants of 126bp HVS-2 (nt 34-159) D-Loop mtDNA in the male-female siblings of Madurese Population (N: 17)

	Nucleotide (nt)												
	16270	16286	16290	16310	16343	16367	16387	16390	16396	16398	16400	16401	16409
rCRS	C	C	C	A	A	A	A	G	T	T	G	T	A
Male	G	T	T	A	T	G	C	T	T	A	G	T	A
Male Siblings	A	T	A	G	A	C	A	C	G	T	T	A	G
%	3%	5%	8%	8%	9,5 %	11%	5%	6%	10%	7,5 %	10%	7%	10%

Table 6. The variants of 126bp HVS-2 (nt 34-159) D-Loop mtDNA in the male-male siblings of Madurese Population (N: 13)

	Nucleotide (nt)										
	16280	16289	16299	16310	16377	16387	16393	16398	16403	16406	16408
rCRS	A	T	A	G	C	A	C	G	C	T	C
Male	T	G	T	A	A	C	T	T	T	A	G
Male Siblings	A	T	C	A	A	T	A	C	C	T	T
%	5%	10%	9,5 %	8,5 %	8%	10%	11,5 %	7,5 %	10,5 %	9,5 %	10%

The homological analysis of female-female siblings indicates 11 variants or morphs on 143bp HVS-1 D-Loop mtDNA. The variants with the highest percentage are (16387A→G, 16387A→C: 15%) in female-female siblings and (16393C→T, 16393C→A: 11.5%) in male-female siblings. Meanwhile, the homological analysis of male-female siblings indicates 13 variants on 143bp HVS-1 D-Loop mtDNA (nt 16268-16410), with the highest percentage on

the 16367A→G, 16367A→C: 11% variant.

4. Discussion

The findings of this study showed 11 variants of 126bp HVS-2 D-Loop mtDNA nt 34-159 in female-female siblings, male-female siblings, and male-male siblings samples of the Madurese population. 11 variants were also found on 143bp HVS-1 D-Loop mtDNA nt 16268-16410 in female-female and male-male siblings' samples of the Madurese population. Meanwhile, 13 variants of 143bp HVS-1 D-Loop mtDNA were found in male-female sibling samples of the Madurese population. These variants are specific to the Madurese population living on the island of Madura because only a few studies on the variants of D-Loop region nucleotides have been conducted recently, particularly on the 26bp HVS-2 D-Loop mtDNA nt 34-159 and 143bp HVS-1 D-Loop mtDNA nt 16268-16410 loci.

In general, the Madurese population can be grouped into two major groups, namely those who live on the island of Madura and those who migrate to the northern coasts and the *horseshoe* regions of the islands of Java. Although most of the areas included in the Madura regions consist of islands, the Madurese population has its own unique culture. Since ancient times, the Madurese people have dared to migrate, leaving their home island. This is proven that the Madurese people have spread across the country. They can even be found in other countries [6].

Population groupings occur during the migration due to waves of people migrating from one place to another. The consequence of the migration waves is interbreeding (i.e., marriage between population groups), leading to the mixing genetic materials. There is a possibility that the future population may dominate and inherits several traits from the previous population [11,12]. In this study, the volunteers are those of pure Madurese and born in Madurese regions and adopting endogamous marriage by marrying fellow Madurese population. However, due to the development of technology and transportation, the endogamous marriage rate tends to decrease, as proven by many nucleotide variants found in this study. In a study on population, a clear understanding of sampling processes is essential in obtaining the accuracy of DNA donors and their ancestral genealogy [12].

Four significant mechanisms can change the frequency of genes and genotypes in a population. They are mutation, selection, gene flow, and genetic drift. Mutation and gene flow can increase the variations inside a population. Gene flow is the exchange of genetic materials between two populations caused by migration and marriage processes. Further analysis needs to be conducted on the findings of this study to analyze whether the variants found in this study are included in specific haplotypes or haplogroups [13].

Genetic drift happens in populations living in secluded or isolated areas with a small number of migrations. Bottleneck and founder effect are the two conditions that may trigger genetic drift. A bottleneck is a condition where the size of a population decreases at a certain period; meanwhile, the founder effect is a condition that all individuals in a population originate from a small number of individuals in the population [11, 12, 13]. The findings of this study show that the highest percentage of 126bp HVS-2 variants is (120C→A: 11.5%) in male-female siblings and (131T→C: 11.5%) in male-male siblings. On the other hand, the percentages of 143bp HVS-1 variants are (16387A→G, A→C: 15%) in female-female siblings, (16393C→T, C→A: 11.5%) in male-female siblings, and (16367A→G, A→C: 11%) in male-

male siblings. Different base sequences found in individuals of the same species are genetic markers. Two individuals sharing the same genetic markers at the same position indicate that these individuals have kinship relationships. From this genetic marker, we can trace the origins of our ancestors and where they came from. The more specific genetic markers possessed by individuals in a race or species, the more diverse the characteristics of the individuals [13,14].

Genetic diversity decreases because of migrations. When a small group of our ancestors migrated to a new area, they brought a small portion of the genetic diversities of their original community [13,15].

Classical theories perceive human evolution as the effects of external factors such as climate, geography, and topology. The latest perspectives of anthropology state that cultural changes may be one of the potential factors that change the environment and habits, which triggers a rapid evolution resulting from the interaction between genetics and culture [13,15].

The findings of this study recommend that studies on population should not only focus on the genetic aspects. Future studies should also consider the cultural factors and customs of a population. A gene pool is not only a group of genes but also an organized dynamic system that contains the history of a population [9,14.15]. Every genetic information has its historical, anthropological, and statistical aspects; therefore, coordination and collaboration of various fields of studies are essential.

5. Conclusion

This study has successfully analyzed the fragments of mtDNA sized 126bp D-Loop HVS-2 nt 59-134 and 143bp D-Loop HVS-1 nt 16268-16410 of siblings from the Madurese population. The results of homological analysis on 126bp HVS-2 find 11 variants in female-female, male-female, and male-male siblings. Meanwhile, a homological study on 143bp HVS-1 finds 11 variants in female-female and male-female siblings and 13 variants in male-male siblings. The variants with the highest percentage are 129G→C, 16387A→C, and 16387A→G, with a percentage of 15%.

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