J Genet Resour2021;7(2): 220-226 *Homepage: http://sc.journals.umz.ac.ir/*

RESEARCH ARTICLE *DOI: 10.22080/jgr.2021.20887.1239*

J|G|R

Forensic Genetic Analysis of Mitochondrial DNA Hypervariable Region III Sequences in Muslims from South India

Somayyeh Samehsalari[1] and Adimoolam Chandrasekar[2]

[1] Department of Social Science, University of Mazandaran, Babolsar, Mazandaran, Iran
[2] Anthropological Survey of India, Southern Regional Centre, Bogadi, Mysore, India

ARTICLE INFO

Article history:
Received 26 March 2021
Accepted 30 May 2021
Available online 07 June 2021

Keywords:
Mitochondrial DNA
Hypervariable region III
Forensic analysis
Muslim population
India

Corresponding authors:
✉ S. Samehsalari
salarisonya@gmail.com

p-ISSN 2423-4257
e-ISSN 2588-2589

ABSTRACT

The absence of recombination, high rate of mutation, and maternal inheritance of the mitochondrial DNA (mtDNA) genome makes it a valuable tool in many fields including evolutionary anthropology, genetic genealogy, population history, and forensic science. The mtDNA genome can be separated into two parts: a large coding region and a smaller fragment called the control region or non-coding region that generally referred to as displacement loop (D loop). The mtDNA D-loop region was highly polymorphic and has proven a precious marker in forensic identification. The study aims to examine and suggests polymorphism of the HVRIII region as a power marker along with HVRI and HVRII in forensic investigations. Within the control region of the mtDNA genome, the sequences of hypervariable region III (HVR III) (nucleotide position 438-574) were obtained from 60 unrelated Muslims of Shrirangapattana town, located in Karnataka state of South India. The complete mtDNA control region was amplified and sequenced by the Sanger sequencing method. The study provided the identification of 18 different haplotypes and 17 polymorphic nucleotide positions. The most common haplotype (H.18) was consistent with the Anderson sequence which occurred fourteen times. The distribution of nucleotide substitutions, insertions, and deletions was computed and determined that transitions made up the majority of the variations (58%) in this region. The genetic diversity was estimated at 0.89939 and the random match probability at 0.1155. The power of discrimination was found to be 0.8844 and the rest of the statistical parameters such as the mean of pair-wise differences and nucleotide diversity were established as 2.255932 ± 1.25884 and 0.010071± 0.00623, respectively. Consequently, the discovery of high genetic, haplotype, and nucleotide diversity, and high power of discrimination impart the use of hypervariable region III (HVR III) as an important marker in forensic investigations.

Please cite this paper as: Samehsalari S, Chandrasekar A. 2021. Forensic genetic analysis of mitochondrial DNA hypervariable region III sequences in Muslims from South India**.** *J Genet Resour* 7(2): 220-226. doi: 10.22080/jgr.2021.20887.1239.

Introduction

The mtDNA sequence analysis in the D-loop region has found a useful tool in forensic investigations and matrilineal origin research, especially when there is insufficient nuclear DNA in samples for typing (Hwa *et al*., 2010; Panneerchelvam *et al*., 2010). The absence of recombination, high copy number, and circular form of mtDNA make it a valuable marker in identifying persons from the highly degraded biological sample in the human identification field (Fridman and Gonzalez, 2009). Although the most extensive mtDNA variations between individuals are found within the two segments of control region hypervariable region I (HVRI) (np 16024 to 16365) and hypervariable region II (HVRII) (np 73 to 340), occasionally a third portion of the control region, known as

hypervariable region III (HVRIII) (np 438 to 574) can provide evidence with the identity of crime victims (Bini *et al*., 2003; Mabuchi *et al*., 2007) especially when sequences of HVRI and HVRII are identical, the additional polymorphic site within HVRIII can be helpful to identification such indistinguishable HVRI/HVRII samples (Baasner *et al*., 1998; Lutz *et al*., 2000; Budowle *et al*., 2003). Different investigations have been shown the potential value of discrimination and identification using polymorphic sites in HVRI and HVRII regions (Imaizumi *et al*., 2002; Bini *et al*., 2003; Gabriel *et al*., 2003; Divne *et al*., 2005; Tsutsumi *et al*., 2006; Mabuchi *et al*., 2007; Lander *et al*., 2008; Lehocký *et al*., 2008; Fridman and Gonzalez, 2009); however, only a few groups have been working with HVRIII analysis for this purpose (Andrews *et al*., 1999; Lutz *et al*., 2000; Nagai *et al*., 2004; Lee *et al*., 2006; Hameed and Jebor, 2016). In this study, we evaluated the variability of the HVRIII region in the Indian Muslim population to find polymorphic positions that fulfill the situations necessary for their future application in the identification process and forensic investigation casework**.**

Materials and methods

Population and DNA extraction

After obtaining written consent, which was approved by the ethical committee of the University of Mysore, blood samples were obtained from 60 randomly chosen unrelated Indian Muslim individuals of Shrirangapattana. From each 5ml of intravenous blood was collected in 6 ml EDTA-K2 tubes (BD Vacutainer). The blood samples were subjected to DNA extraction using a DNA extraction kit, QIAamp DNA mini kit (Qiagen, Hilden, Germany) according to the manufacturer's instructions. From the extracted DNA the targeted mtDNA region was amplified using the polymerase chain reaction (PCR) technique with the help of specified primers and sequences obtained by using Sanger's method. Mutations of these sequences were scored after comparing with the Revised Cambridge Reference Sequence and further analyzed by using an appropriate statistical tool.

PCR amplification and sequencing

For PCR amplification of the HVRIII region of mtDNA, the following primers were used: HV3F (5'-GAGCCCGTCTAAACATTTTCAG) and HV3R (5'-CAGCACTTAAACACATCTCTGC). PCR amplification was carried out in 40 µl reaction mixture subjected to 35 cycles of 95 °C for 3 min, 95 °C for 30 s, 58 °C for 30 s, 72 °C for 45 s, and a final extension at 72 °C for 10 min. The DNA sequencing was carried out in Agrigenome labs (A subsidiary of Scigenom labs) using HV3F forward and HV3R reverse primers by Sanger's method.

Statistical Analysis

The sequences of HVRIII (position 438-574) aligned and compared with the Revised Cambridge Reference Sequence (rCRS) (Andrews *et al*., 1999) using clustal W software in the bioEdit version 7.2.6. The probability of randomly selecting two individuals with the same mitochondrial profile [random match probability ($_P$)] was defined by an equation $P=\sum X^2$, where X is the frequency of each observed mtDNA haplotype (Stoneking *et al*., 1991), The genetic diversity (h) was calculated following $h=n(1-\sum X^2)/ (n-1)$, where n is the sample size and X is the frequency for each observed haplotype, and The discrimination power (DP) was also estimated using the equation $DP= (1-\sum X^2)$, where X is the frequency of each observed haplotype (Tajima, 1989). The rest of the statistical parameters were calculated using Arlequin version 3.5.2 software (Excoffier and Lischer, 2010).

Results and discussion

After performance sequence alignment against the Revised Cambridge Reference Sequence, variations from the rCRS represented mutations, nucleotide positions of these mutations were determined and subsequently, mitochondrial haplotypes were concluded and correlated to the published databases. To investigate the HVRIII sequences from nucleotide position 438 to 574, the data were categorized into nucleotide substitutions, deletions, and insertions. Seventeen different variable sites of HVRIII were observed, as shown in table 1.

The most frequently observed nucleotide substitution was at position 489 (T to C) and secondly at position 523 (A deletion) and 524 (C deletion). T to C transition at np 489 is the most frequent variant in the human population. In the present study, it is observed to be 63.34 percent. Similar high polymorphism at np 489 found in Thai populations (66.67%) (Thongngam *et al*., 2016), Japanese (64.67%) (Nagai *et al*., 2004) and (64.3%) (Tsustumi *et al*., 2006), Koreans (61.82%) (Zhang *et al*., 2005), Han Chinese (56.86) (Zhang YJ *et al*., 2005), Hong Kong people (50.93%) (Irwin *et al*., 2009), and Malaysians (45.16%) (Nur Haslindawaty *et al*., 2010), while T489C nucleotide variation in Iraqis (4.8%) (Shneewer *et al*., 2015), Venezuelans (29%) (Lander *et al*., 2008), and Germans (6.5%) (Lutz *et al*., 1998) showed lower polymorphism. This may be region-specific to Southeast Asian populations. C to G at np 447 characterizes haplogroup M2. M2 haplogroup is specific to Indian (Chandrasekar *et al*., 2009). In this study, it forms the H3 haplotype with a frequency of about 12 percent. Transition C to T at np 461 is specific to the M6 haplogroup and Southeast Asia, particularly the Indian subcontinent. The next variant at np 462 is more frequent in the mtDNA database and it is nested with J1b, J1c, and J1d haplogroup trees. It probably originated in the near east and frequent in Pakistan (Quintana *et al*., 2004). Transition T-C at np 480 is frequent in European lineages whereas the same at np 482 is frequent in Indian populations. G499A is about 6 percent in the mitomap database and frequent in the American continent (B2 haplogroup). Haplotype H12, A508G nested with U2e lineage which is frequent in Italy (Achilli *et al*., 2007). In the present study, it forms haplotype H6.

Table 1. Sequence polymorphisms in the mtDNA HVRIII region of 60 unrelated Indian Muslims.

Anderson	447C	461C	462C	480T	482T	489T	499G	508A	511C	523A	524C	524a*	524b*	524c*	524d*	573.1	574A	Number of Individual
H*1	.	.	.	.	.	C	.	.	T	.	.	.	.	.	.	.	.	2
H2	.	.	.	.	.	C	.	.	.	.	.	.	.	.	.	.	.	9
H3	G	.	.	.	.	C	.	.	.	D	D	.	.	.	.	.	.	7
H4	.	.	T	.	.	C	.	.	.	.	.	.	.	.	.	.	.	5
H5	.	.	.	.	.	C	.	.	.	D	D	.	.	.	.	.	.	2
H6	.	.	.	.	.	.	.	G	.	.	.	.	.	.	.	.	.	1
H7	.	.	.	.	.	C	.	.	.	.	.	.	.	.	.	.	C	1
H8	.	.	.	.	.	.	.	.	.	D	D	.	.	.	.	.	.	4
H9	.	.	.	.	C	C	.	.	.	D	D	.	.	.	.	.	.	2
H10	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	C	.	1
H11	.	.	.	.	C	C	.	.	.	.	.	.	.	.	.	.	.	5
H12	.	.	.	.	.	.	A	.	.	.	.	.	.	.	.	.	C	1
H13	.	.	.	.	.	C	.	.	.	.	.	A	C	.	.	.	.	1
H14	.	.	.	.	.	C	.	.	.	.	.	A	C	A	C	.	.	1
H15	.	.	.	C	.	.	.	.	.	.	.	.	.	.	.	.	.	1
H16	.	T	.	.	.	C	.	.	.	D	D	.	.	.	.	.	.	2
H17	.	.	.	.	C	C	.	.	.	.	.	.	.	.	.	C	.	1
H18	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	14
Total	.	.	.	.	.	12	.	.	.	.	.	.	.	.	.	.	.	60

H*: haplotype; G: Guanine; T: Thiamine; C: Cytosine; A: Adenine; D: deletion; (a-d)*: indicate additions of nucleotides not found in the reference sequence.

From HVRIII region of mtDNA, 18 different haplotypes were identified (Table 2). Among them, 8 types were observed only once and the other 10 haplotypes were shared between multiple individuals; 4 types were observed twice. The haplotype no.18 was the most frequently detected haplotype, which is the reference sequence (rCRS). Reported variable sequences in this manuscript are deposited in the Genbank under accession number MK104528-MK104587.

Table 2. mtDNA HVRIII haplotypes among the 60 unrelated Indian Muslims.

No	HVRIII-haplotype (438-574)	Frequency	Percentage
H*1	489, 511	2	0.0333
H2	489	9	0.1500
H3	447, 489, 523 D, 524 D	7	0.1167
H4	462, 489	5	0.0833
H5	489, 523 D, 524 D	2	0.0333
H6	508	1	0.0167
H7	523 D, 524 D	4	0.0667
H8	482, 489	5	0.0833
H9	499, 574	1	0.0167
H10	489, 524. 1 A, 524.1 C	1	0.0167
H11	489, 524.2 A, 524.2 C	1	0.0167
H12	482, 489, 523 D, 524 D	2	0.0333
H13	480	1	0.0167
H14	461, 489, 523 D, 524 D	2	0.0333
H15	489, 574	1	0.0167
H16	482, 489, 573.1 C	1	0.0167
H17	573.1 C	1	0.0167
H18	rCRS	14	0.2333
	Total	60	1

H*: haplotype; D: deletion; 0.1, 0.2: Insertion; rCRS: Revised Cambridge Reference Sequence.

Table 3 exhibits the frequency distribution of nucleotide substitutions, insertions, and deletions. It was observed that transitions (58%) were more prevalent than transversions (14%). It is also observed that deletions were more than insertions in the HVRIII region of mtDNA. Nevertheless, the HVR III has proven to be useful as a supplementary polymorphic segment for forensic application (Hoong and Lek, 2005). Even though the majority of forensic cases utilize only the HVR I and HVR II sequences, it is noteworthy that HVR III $(AC)_n$ repeat segment has equal potential as an additional mtDNA marker in forensic investigations (Ivanov *et al*., 1996; Lutz *et al*., 2000; Chung *et al*., 2005; Fridman and Gonzalez, 2009; Bhatti *et al*., 2018). The nucleotide insertion was observed in the present study, at two positions with +C at position 573, +AC at position 524, and + ACAC at position 524. Nucleotide deletion was also observed at nucleotide position 522-523 was a deletion of -(AC), which was found in 5 of 18 individuals in our study. $(CA)_n$ dinucleotide indels of mtDNA HVR III 514-524 region in the Indian population has been found in the Urali Kuruman tribe of Kerala, South India. Among the Uralikuruman, CA_4 is the common one. (Sylvester *et al*., 2018). It was also found that CA_4 was common among the Thai population (Thongngam *et al*., 2016) whereas the common CA repeat observation in the present studied population was CA_5.

Table 3. Nucleotide substitutions, insertions, and deletions in the mitochondrial DNA hypervariable HVRIII region of 60 unrelated Indian Muslims.

Mutation type	Number of positions	Total number of mutations
Transition		
C → T	3	9
T → C	3	39
A → G	1	1
G → A	1	1
Total	8	50
Transversion		
C→ G	1	7
A → C	1	2
Total	2	9
Insertion		
+1AC (at np	1	1
524)	1	1
+2AC (at np		
524)		
+C	1	2
Total	3	4
Deletion		
-AC	1	17
Total	1	17

Diversity parameters for the HVRIII region of mtDNA in Indian Muslims of the present study are given in Table 4. A statistical estimate of the results for this population showed a genetic diversity (h) of 0.89939 and it was calculated between 0.6 to 0.8 by several authors (Nagai *et al*., 2004; Vanecek *et al*., 2004; Lee *et al*., 2006). The probability of random match (p) of two individuals showing the same mtDNA

haplotype was 0.1155 and it was defined between the ranges of 0.2 to 0.4 in different human population studies (Bini *et al*., 2003; Nagai *et al*., 2004; Vanecek *et al*., 2004; Lee *et al*., 2006; Palencia *et al*., 2010). Similar to the present work high genetic diversity (h) 0.860 for the HVR-III region and the power (1–p) of discrimination 0.846 for HVR-III among the Tai populations was found (Chen *et al*., 2002).

In this study, the power of calculated discrimination was 0.8844, and statistical parameters such as nucleotide diversity and mean the number of pair-wise differences was estimated as 0.010071± 0.006230 and 2.2555932± 10258400 respectively. Thus, the high genetic diversity for the HVRIII region in the present studied population was found.

Table 4. Diversity measures for 60 unrelated Indian Muslims.

Parameters	HVIII
Genetic diversity (h)	0.89939
Random match probability (P)	0.1155
Nucleotide diversity(π_n)	0.010071 +/- 0.006230
Mean number of pairwise differences (π)	2.255932 +/- 1.258400
Power of discrimination (PD)	0.8844

Conclusion

Sequence databases are the best source of information regarding the power of mtDNA for identity testing. We have examined the nucleotide diversity of the HVRIII region in 60 unrelated individuals of a socially and culturally distinct group. A total of seventeen variations were observed in the 137 bp length stretch of the HVRIII region. The nucleotide transitions are found to be more predominant than the transversions (50:9). T to C transition was found to be the most frequent substitution (66%). The present studied population has CA_5 type of CA repeat as the most common type. It reveals that the present population has more genetic relation with European populations. The high diversity (0.8994), power of discrimination (0.8844) for the HVRIII region of the studied population is in agreement with the results of similar studies. Although combined HVRI, HVRII, and HVRIII data (D loop) together by having more variable site and more haplotypes increase the power of discrimination, identification, and the genetic diversity of any population but the additional polymorphic sites within the HVRIII region help to resolve indistinguishable HVRI/HVRII samples and can be used as an additional marker in genetic differentiation of human populations and forensic investigation casework.

Acknowledgments

The authors are gratefully acknowledging Dr. G.B. Aravind (Associate Professor in Department of Forensic Medicine & Toxicology, JSS Medical College, Mysore) for constructive discussions and excellent support in the fieldwork.

Conflicts of interest

The authors declare that they have no conflict of interest.

References

Achilli A, Olivieri A, Pala M, Metspalu E, Fornarino S, Battaglia V, Torroni A. 2007. Mitochondrial DNA variation of modern Tuscans supports the near eastern origin of Etruscans. *Am J Hum Genet* 80: 759-768.

Andrews RM, Kubacka I, Chinnery PF, Lightowlers RN, Turnbull DM, Howell N. 1999. Reanalysis and revision of the Cambridge reference sequence for human mitochondrial DNA. *Nat Genet* 23: 147.

Baasner A, Schäfer C, Junge A, Madea B. 1998. Polymorphic sites in human mitochondrial DNA control region sequences: population data and maternal inheritance. *Forensic Sci Int* 98: 169-178.

Bhatti S, Aslam Khan M, Abbas S, Attimonelli M, Gonzalez GR, Aydin HH, de Souza EM. 2018. Problems in mitochondrial DNA forensics: while interpreting length heteroplasmy conundrum of various Sindhi and Baluchi ethnic groups of Pakistan. *Mitochondrial DNA Part A* 29: 501-510.

Bini C, Ceccardi S, Luiselli D, Ferri G, Pelotti S, Colalongo C, Falconi M, Pappalardo G. 2003. Different informativeness of the three hypervariable mitochondrial DNA regions in the population of Bologna (Italy). *Forensic Sci Int* 135: 48-52.

Budowle B, Allard MW, Wilson MR, Chakraborty R. 2003. Forensics and mitochondrial DNA: applications, debates,

and foundations. *Annu Rev Genomics Hum Genet* 4: 119-141.

Chandrasekar A, Kumar S, Sreenath J, Sarkar BN, Urade BP, Mallick S, Kiran U. 2009. Updating phylogeny of mitochondrial DNA macrohaplogroup m in India: dispersal of modern human in South Asian corridor. *Plos One* 4: e7447

Chen MH, Lee HM, Tzen CY. 2002. Polymorphism and heteroplasmy of mitochondrial DNA in the D-loop region in Taiwanese. *J Formos Med Assoc* 101(4): 268-276.

Chung U, Lee HY, Yoo JE, Park MJ, Shin KJ. 2005. Mitochondrial DNA CA dinucleotide repeats in Koreans: the presence of length heteroplasmy. *Int J Legal Med* 119: 50-53

Divne AM, Nilsson M, Calloway C, Reynolds R, Erlich H, Allen M. 2005. Forensic casework analysis using the HVI/HVII mtDNA linear array assay. *J Forensic Sci* 50: 1-7.

Excoffier L, Lischer HE. 2010. Arlequin suite ver 3.5: a new series of programs to perform population genetics analyses under Linux and Windows. *Mol Ecol Resour* 10: 564-567.

Fridman C, Gonzalez RS. 2009. HVIII discrimination power to distinguish HVI and HVII common sequences. *Forensic Sci Int Genet Suppl Ser* 2: 320-321.

Gabriel MN, Calloway CD, Reynolds RL, Primorac D. 2003. Identification of human remains by immobilized sequence-specific oligonucleotide probe analysis of mtDNA hypervariable regions I and II. *Croat Med J* 44: 293-298.

Hameed IH, Jebor MA. 2016. Forensic analysis of mitochondrial dna hypervariable region HVII (encompassing nucleotide positions 37 to 340) and HVIII (encompassing nucleotide positions 438-574) and evaluating the importance of these variable positions for forensic genetic purposes. *J Univ Babylon* 24: 1246-1259.

Hoong LL, Lek KC. 2005. Genetic polymorphisms in mitochondrial DNA hypervariable regions I, II and III of the Malaysian population. *Asia Pacific J Mol Biol Biotechnol* 13: 79-85

Hwa HL, Ko TM, Chen YC, Lin CY, Huang YH, Tseng LH, Lee JC. 2012. Sequence polymorphisms of mtDNA HV1, HV2 and HV3 regions in eight population groups living in Taiwan. *Aust J Forensic Sci* 44: 243-252.

Imaizumi K, Parsons TJ, Yoshino M, Holland MM. 2002. A new database of mitochondrial DNA hypervariable regions I and II sequences from 162 Japanese individuals. *Int J Legal Med* 116: 68-73.

Irwin JA, Saunier JL, Beh P, Strouss KM, Paintner CD, Parsons TJ. 2009. Mitochondrial DNA control region variation in a population sample from Hong Kong, China. *Forensic Sci Int Genet* 3: 119-125.

Ivanov PL, Wadhams MJ, Roby RK, Holland MM, Weedn VW, Parsons TJ. 1996. Mitochondrial DNA sequence heteroplasmy in the Grand Duke of Russia Georgij Romanov establishes the authenticity of the remains of Tsar Nicholas II. *Nature Genet* 12: 417-420.

Lander N, Rojas MG, Chiurillo MA, Ramírez JL. 2008. Haplotype diversity in human mitochondrial DNA hypervariable regions I–III in the city of Caracas (Venezuela). *Forensic Sci Int Genet* 2: 61-64.

Lee HY, Yoo JE, Park MJ, Chung U, Shin KJ. 2006. Mitochondrial DNA control region sequences in Koreans: identification of useful variable sites and phylogenetic analysis for mtDNA data quality control. *Int J Legal Med* 120: 5-14.

Lehocký I, Baldovič M, Kádaši Ľ, Metspalu E. 2008. A database of mitochondrial DNA hypervariable regions I and II sequences of individuals from Slovakia. *Forensic Sci Int Genet* 2: e53-59.

Lutz S, Wittig H, Weisser HJ, Heizmann J, Junge A, Dimo-Simonin N et al., Parson W, Edelmann J, Anslinger K, Jung S, Augustin C. 2000. Is it possible to differentiate mtDNA by means of HVIII in samples that cannot be distinguished by sequencing the HVI and HVII regions? *Forensic Sci Int* 113: 97-101.

Lutz S, Weisser HJ, Heizmann J, Pollak S. 1998. Location and frequency of polymorphic positions in the mtDNA control region of individuals from Germany. *Int J Leg Med* 111: 67-77.

Mabuchi T, Susukida R, Kido A, Oya M. 2007. Typing the 1.1 kb control region of human

mitochondrial DNA in Japanese individuals. *J Forensic Sci* 52: 355-363.

Nagai A, Nakamura I, Bunai Y. 2004. Sequence analysis of mitochondrial DNA HVIII region in a Japanese population. *Int Congr Elsevier* 1261: 410-412.

Nur Haslindawaty AR, Panneerchelvam S, Edinur HA, Norazmi MN, Zafarina Z. 2010. Sequence polymorphisms of mtDNA HV1, HV2, and HV3 regions in the Malay population of Peninsular Malaysia. *Int J Leg Med* 124: 415-426.

Palencia L, Valverde L, Álvarez A, Cainé LM, Cardoso S, Alfonso-Sánchez MA, Pinheiro MF, de Pancorbo MM. 2010. Mitochondrial DNA diversity in a population from Santa Catarina (Brazil): predominance of the European input. *Int J Legal Med* 124: 331-336.

Panneerchelvam S, Edinur HA, Norazmi MN, Zafarina Z. 2010. Sequence polymorphisms of mtDNA HV1, HV2, and HV3 regions in the Malay population of Peninsular Malaysia. *Int J Legal Med* 124: 415-426.

Quintana ML, Chaix R, Wells RS, Behar DM, Sayar H, Scozzari R, Rengo C, Al-Zahery N, Semino O, Santachiara-Benerecetti AS, Coppa A. 2004. Where west meets east: the complex mtDNA landscape of the southwest and Central Asian corridor. *Am J Hum Genet.* 74: 827-845.

Shneewer HA, Al-loza NG, Kareem MA, Hameed IH. 2015. Sequence analysis of mitochondrial DNA hypervariable region III of 400 Iraqi volunteers. *Afr J Biotechnol* 14: 2149-2156.

Stoneking M, Hedgecock D, Higuchi RG, Vigilant L, Erlich HA. 1991. Population variation of human mtDNA control region sequences detected by enzymatic amplification and sequence-specific oligonucleotide probes. *Am J Hum Genet* 48: 370.

Sylvester C, Krishna MS, Rao, JS, Chandrasekar A. 2018. Allele frequencies of mitochondrial DNA HVR III 514-524 $(CA)_n$ dinucleotide repeats in the Urali Kuruman tribal population of South India. *Egypt J Forensic Sci* 8: 1-5.

Tajima F. 1989. Statistical method for testing the neutral mutation hypothesis by DNA polymorphism. *Genetics* 123: 585-595.

Thongngam P, Leewattanapasuk W, Bhoopat T, Sangthong P. 2016. Nucleotide sequence analysis of the hypervariable region III of mitochondrial DNA in Thais. *J Forensic Leg Med* 41: 10-14.

Tsutsumi H, Komuro T, Mukoyama R, Nogami H. 2006. Hypervariable region structure and polymorphism of mtDNA from dental pulp and a family analysis. *J Oral Sci* 48: 145-152.

Vanecek T, Vorel F, Sip M. 2004. Mitochondrial DNA D-loop hypervariable regions: Czech population data. *Int J Legal Med* 118: 14-18.

Zhang Y, Xu Q, Cui H, Cui Y, Lin H, Kim K, et al. 2005. Haplotype diversity in mitochondrial DNA hypervariable region I, II and III in a Korean ethnic group from northeast China. *Forensic Sci Int* 151: 299-301.

Zhang YJ, Xu QS, Zheng ZJ, Lin HY, Lee J Bin. 2005. Haplotype diversity in mitochondrial DNA hypervariable region I, II and III in northeast China Han. *Forensic Sci Int* 149: 267-269.

J Genet Resour2021;7(2): 220-226 Homepage: http://sc.journals.umz.ac.ir/
RESEARCH ARTICLE DOI: 10.22080/jgr.2021.20887.1239
J|G|R

تجزیه و تحلیل ژنتیک پزشکی قانونی توالی‌های ناحیه سوم HVRIII از ژنوم میتوکندری در مسلمانان جنوب هند

سمیه سامه سالاری[1]، آدیمولام چاندراسکار[2]

[1]گروه علوم اجتماعی ، دانشگاه مازندران ، بابلسر ، مازندران ، ایران

[2]مرکز تحقیقات انسان شناسی جنوب هند، بوگادی، میسور، هندوستان

چکیده

فقدان نوترکیبی، میزان بالای نرخ جهش و وراثت مادری ژنوم میتوکندری آن را به یک ابزار ارزشمند در بسیاری از زمینه‌ها مانند انسان شناسی تکاملی، شجره نامه ژنتیکی، تاریخچه جمعیت و پزشکی قانونی تبدیل کرده است. ژنوم میتوکندری را می‌توان به دو بخش مجزا تقسیم کرد: یک ناحیه بزرگ رمزشونده و یک قطعه کوچکتر به نام ناحیه کنترل و غیر رمز شونده که به طور کلی به عنوان D-loop شناخته می‌شود. ناحیه D-loop از ژنوم میتوکندری بسیار متغیر است و به عنوان یک نشانگر با ارزش در تعیین هویت در پزشکی قانونی ثابت شده است. هدف از این مطالعه بررسی میزان تغییرات ناحیه HVRIII و کاربرد این ناحیه همراه با نواحی HVRI و HVRII در تحقیقات پزشکی قانونی می‌باشد .در ناحیه کنترل ژنوم میتوکندری، توالی ناحیه سوم HVIII (جایگاه نوکلئوتیدی ۴۳۸-۵۷۴) از ۶۰ مسلمان غیر خویشاوند شهر Shrirangapattana، واقع در ایالت کارناتاکا در جنوب هند ، بدست آمد. ناحیه کنترل ژنوم میتوکندری به صورت کامل با روش تعیین توالی Sanger توالی یابی گردید. این مطالعه، شناسایی ۱۸ هاپلوتیپ مختلف و ۱۷ جایگاه نوکلئوتیدی متغیر را فراهم کرده است. رایج ترین هاپلوتیپ (H.18) با توالی اندرسون (مرجع) مطابقت داشت و چهارده بار رخ داده بود. توزیع انواع نوکلئوتید های جهش یافته insertions و deletions محاسبه و مشخص شد که transitions بیشترین تغییرات (۵۸٪) را در این ناحیه ایجاد کردند. تنوع ژنتیکی ۰/۸۹۹۳۹و احتمال توالی‌های کاملا مشابه ۰/۱۱۵۵ برآورد شد. قدرت تمایز ۰/۸۸۴۴ بدست آمد و بقیه پارامترهای آماری مانند میانگین اختلاف جفت نوکلئوتیدی و تنوع نوکلئوتیدی به ترتیب ۱/۲۵۸۸۴±۲/۲۵۵۹۳۲ و ۰/۰۰۶۲۳ ± ۰/۰۱۰۰۷۱ تعیین شد. در نتیجه، تنوع بالای ژنتیکی، هاپلوتیپ و نوکلئوتیدی و قدرت بالای تمایز، استفاده از ناحیه سوم HVIII را به عنوان یک نشانگر مهم در تحقیقات پزشکی قانونی تائید می‌کند.

واژگان کلیدی: ژنوم میتوکندری؛ ناحیه متغیر سوم؛ پزشکی قانونی؛ جمعیت مسلمان؛ هندوستان

Please cite this paper as: Samehsalari S, Chandrasekar A. 2021. Forensic genetic analysis of mitochondrial DNA hypervariable region III sequences in Muslims from South India**.** *J Genet Resour* 7(2): 220-226. doi: 10.22080/jgr.2021.20887.1239.