Scientific Foundation SPIROSKI, Skopje, Republic of Macedonia
Open Access Macedonian Journal of Medical Sciences. 2022 May 27; 10(A):1089-1092.
https://doi.org/10.3889/oamjms.2022.9853
eISSN: 1857-9655
Category: *A - Basic Sciences*
Section: *Genetics*

Genetic Analysis of TPOX, CSF1PO, D3S1358, D8S1179, vWA, D5S818, and TH01 Short Tandem Repeats Loci in Nias Population, Indonesia

Citra Manela*, Taufik Hidayat, Rika Susanti, Noverika Windasari

Department of Forensic Medicine, Faculty of Medicine, Universitas Andalas, Padang, West Sumatra, Indonesia

Edited by: Slavica Hristomanova-Mitkovska
Citation: Manela C, Hidayat T, Susanti R, Windasari N. Genetic Analysis of TPOX, CSF1PO, D3S1358, D8S1179, vWA, D5S818, and TH01 Short Tandem Repeats Loci in Nias Population, Indonesia. Open Access Maced J Med Sci. 2022 May 27; 10(A):1089-1092. https://doi.org/10.3889/oamjms.2022.9853
Keywords: Nias; Short tandem repeats; Loci; Genetic
***Correspondence:** Citra Manela, Department of Forensic Medicine, Faculty of Medicine, Universitas Andalas, Padang, Indonesia. E-mail: manela_84@yahoo.com, citramanela@med.unand.ac.id
Received: 01-May-2022
Revised: 16-May-2022
Accepted: 17-May-2022

Funding: The PNBP fundings of Faculty of Medicine of Universitas Andalas 2021
Competing Interests: The authors have declared that no competing interests exist

Abstract

BACKGROUND: Nias is an island located off the western coast of Sumatra, Indonesia. Nias is situated above the Eurasian and Indo-Australian subduction zone plates. This makes it prone to earthquakes and tsunamis. Genetic analysis and genetic variation of short tandem repeats (STR) locus are not widely known. These data are valuable for individual identification and paternity testing.

METHODS: Seven STR loci (TPOX, CSF1PO, D3S1358, D8S1179, vWA, D5S818, and TH01) were analyzed using 25 healthy and unrelated persons Nias population. Allele frequency, power of discrimination (PD), expected heterozygosity, and probability of exclusion (PE) were calculated.

RESULTS: We found 40 alleles. The allele with highest frequency was alleles 9 at the TH01 loci. While the lowest frequency were allele 9 at the CSF1PO loci, allele 12 at the TPOX loci, alleles 17 and 18 at the D8S1179 loci, and alleles 16 and 20 at the vWA loci. The highest Expected Heterozygosity, PD, and PE at the D8S1179 loci. The highest number of alleles is also at D8S1179 loci. All loci followed the Hardy–Weinberg equilibrium ($p > 0.05$). The PD values for all tested loci ranged from 80.6 to 94.5%.

CONCLUSION: We report the allele frequencies and forensic statistical parameters of seven STR loci (TPOX, CSF1PO, D3S1358, D8S1179, vWA, D5S818, and TH01) in the Nias population, which can be used as a forensic database reference for Nias populations.

Introduction

Microsatellites are a group of molecular markers used for several purposes, including forensic identification, human evolution, and kinship testing [1]. Microsatellites are DNA sequences with varying numbers of short tandem repeats (STR), with repeat units ranging from 2 to 7 bp in length [2], [3]. Microsatellites have been found throughout the genome on all chromosomes. Most microsatellite loci are very small, ranging from a few to a few hundred repeats, and their small size is required for PCR-assisted genotyping. Microsatellites with a higher number of repeats are more polymorphic [4], [5], [6]. The number of repeats at STR loci can vary between individuals.

At each STR locus, there is a statistical likelihood that an individual will have a certain number of repeats [7]. An extensive database known as the combined DNA index system (CODIS) has been set up to process the results of each analysis [8]. The seven loci used in this study are part of the CODIS STR locus.

The Indonesian population consists of various ethnicities and races. The hypervariable locus in many populations has not been well established in Indonesia. Nias is one of the ethnic populations of North Sumatra province in Indonesia. According to the 2020 census, the population of Nias had reached 146.672 [9]. Research conducted by Van Oven *et al.* found that the human population of Nias Island in Indonesia exhibits severely reduced Y chromosome (a non-recombining portion of the Y chromosome [NRY]) and to a lesser extent also reduced mitochondrial DNA (mtDNA) diversity as compared with most other populations from the Asia/Oceania region [10]. There have not been many studies on genetic variation at the STR locus of the Nias tribe. Therefore, this study shows the allele frequency and forensic parameters of the Nias tribe that can be used as a reference for forensic identification and population genetics studies.

Methods

Twenty-five unrelated persons are selected randomly; the sample is a three-generation Nias tribe. Five milliliters of venous blood were taken from the

participant and then put into the EDTA tube. The blood samples were transported to our laboratory at 4°C and stored at−20°C.

To evaluate the performance of the 7 STR loci, we analyze TPOX, CSF1PO, D3S1358, D8S1179, vWA, D5S818, and TH01 STR loci in 25 unrelated Nias populations. Peripheral blood samples were collected from these individuals after acquiring their informed consent. Amplification of 7 loci was performed using GlobalFiler™ Express kit (Thermo Fisher Scientific Company, Carlsbad, USA) in the GeneAmp PCR System 9700 (Thermo Fisher Scientific Company) according to the manufacturer's recommendation. PCR products were separated by capillary electrophoresis in an ABI PRISM 3500 genetic analyzer (Thermo Fisher Scientific Company). The GeneMapper® ID-X software v1.4 (Thermo Fisher Scientific Company) was used for genotype assignment [11], [12].

The allele frequencies were calculated based on genotype counts. The exact tests of Hardy-Weinberg equilibrium were performed using EasyDNA software (https://saasweb.hku.hk/EasyDNA/) [13]. Expected heterozygosity (He), power of discrimination (PD), and probability of exclusion (PE) were also calculated.

This study was conducted by the Ethics Committee of the Faculty of Medicine, Universitas Andalas (No.389/UN.16.2/KEP-FK/2021, date 29.06.2021).

Results

The results showed that there were 40 alleles. At the CSF1PO loci, there were 5 alleles, the highest frequency is allele 10 (0.36). At the TPOX loci, there were 4 alleles, the highest frequency was allele 8 (0.48).

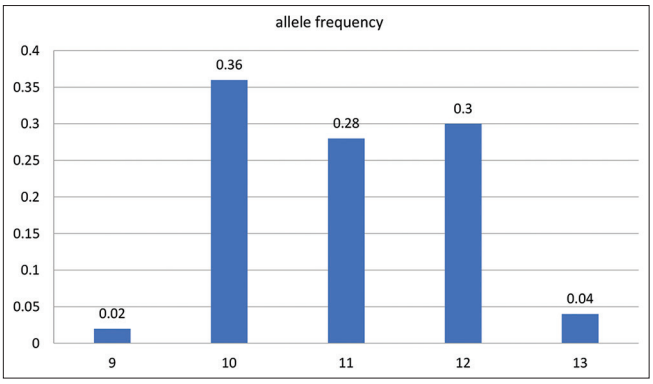

Figure 1: Distribution allele of the CSF1PO loci

The THO1 loci has 7 alleles, with the highest frequency is alleles 9 (0.52). The D3S1358 loci had 4 alleles, the highest frequencies is alleles 16 (0.48). The vWA loci had 7 alleles, the highest frequency is allele 18 (0.46), and the D5S818 loci had 4 alleles

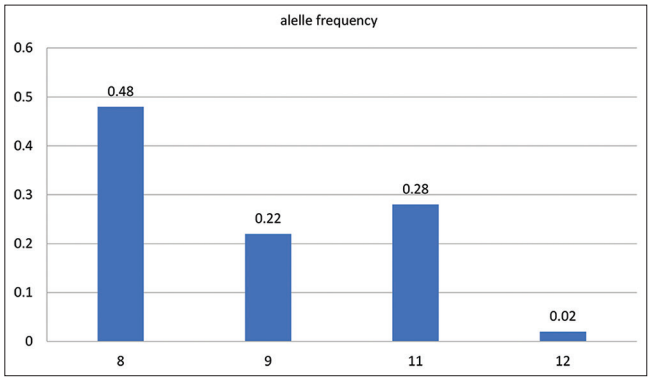

Figure 2: Distribution allele of the TPOX loci

with the highest frequencies is alleles 10 and 11 (0.32) (Figures 1-7).

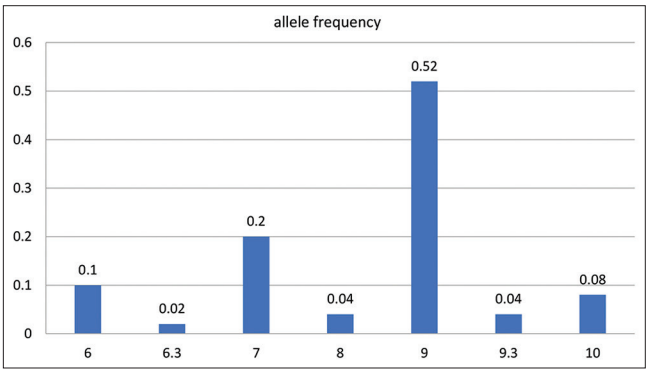

Figure 3: Distribution allele of the TH01 loci

Discussion

There were 40 alleles found at the seven loci used in this study. The allele with the highest frequency was allele 9 at the TH01 loci. The allele with a high frequency indicates that the allele is a common allele found in the Nias population. While the lowest frequency was allele 9 at the CSF1PO loci, allele 12 at the TPOX loci, alleles 17 and 18 at the D8S1179 loci and allele 16 and 20 at the vWA loci The highest number of alleles at the D8S1179 loci was 9 alleles. The lowest number of alleles was seen at the TPOX, D5S818, and D3S1358 loci.

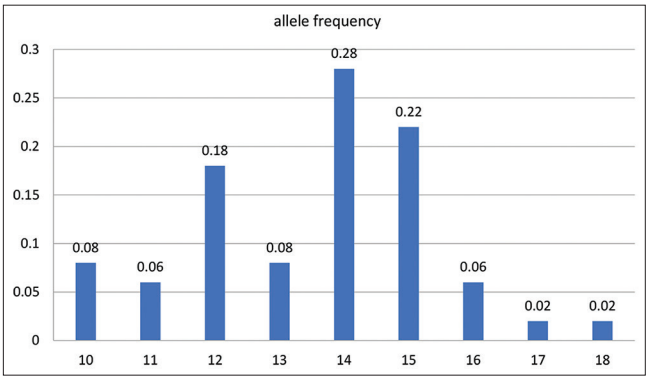

Figure 4: Distribution allele of the D8S1179 loci

Forensic statistical parameters of 7 autosomal STR loci are shown in Table 1. Expected heterozygosity ranged from 0.642 (TPOX) to 0.82 (D8S1179).

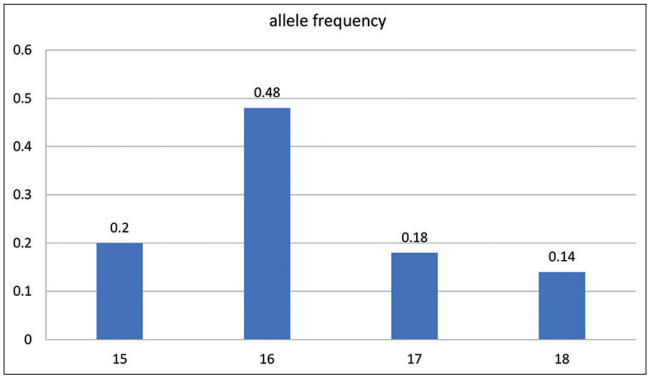

Figure 5: Distribution allele of the D3S1358 loci

According to Butler, the power of exclusion (PE) can be calculated to demonstrate how unusual it is to find a random man who cannot be excluded as the biological father of the child. The PE ranged from 0.346 (TPOX) to 0.65 (D8S1179). The PD for all tested loci ranged from 84.8 to 94.5%. This indicates that these loci can be used to develop a DNA database for the Nias population.

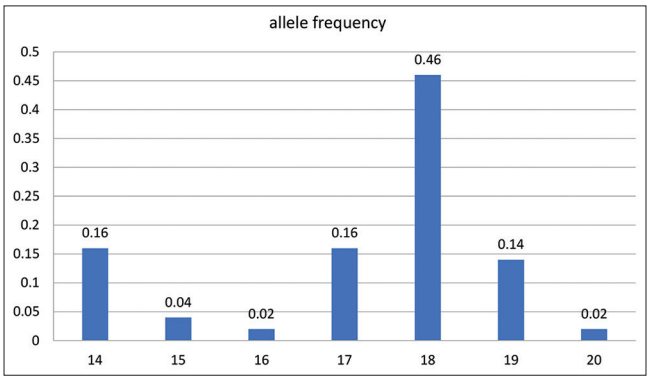

Figure 6: Distribution allele of the vWA loci

The results showed that D8S1179 loci had the highest heterozygosity, power discrimination, and PE from other loci in this study. The highest number of alleles was also found at the D8S1179 loci. The good indicators of the genetic polymorphism within the sample are verified by the number of alleles and the expected heterozygosity in the Nias population. The more number alleles, the higher the value of expected heterozygosity and PD means that the locus is suitable for forensic identification [14].

Table 1: Expected heterozygosity, power of discrimination, probability of exclusion, P value for Hardy–Weinberg equilibrium of TPOX, CSF1PO, D3S1358, D8S1179, VWA, D5S818, and TH01 loci in Nias population

Loci	He	PD	PE	p value for HWE
CSF1PO	0.7	0.851	0.431	1
TPOX	0.642	0.806	0.346	1
TH01	0.67	0.857	0.414	0.832
D8S1179	0.82	0.945	0.65	0.885
D5S818	0.702	0.851	0.432	0.918
D3S1358	0.7678	0.848	0.411	0.41
vWA	0.715	0.884	0.477	0.685

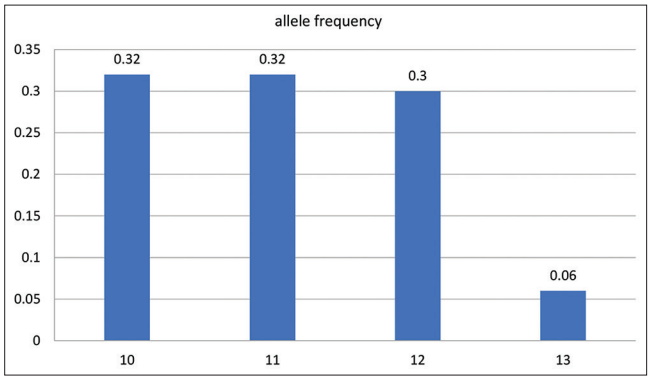

Figure 7: Distribution allele of the D5S818 loci

Conclusion

This study found 40 different alleles from 7 STR loci. Allele 9 at the CSF1PO loci, allele 12 at the TPOX loci, alleles 17 and 18 at the D8S1179 loci, allele 20 at the vWA loci, and allele 11 at the D8S1179 loci had the lowest allele frequencies, while allele 9 at the TH01 loci had the highest allele frequencies. The D8S1179 loci had the highest number of alleles. The loci with the highest heterozygosity were D8S1179, while the locus with the lowest heterozygosity was TPOX. The expected heterozygosity of the 7 STR loci ranged from 64.2% to 82%. The PD for all tested loci ranged from 80.6 to 94.5%. Based on statistical parameters, 7 STR loci in this study can be used for forensic database reference of Nias populations.

References

1. Yamato T, Uchihi R, Nozawa H, Huang XL, Leong YK, Tanaka M, *et al*. Allele distribution at nine STR loc--D3S1358, vWA, FGA, TH01, TPOX, CSF1PO, D5S818, D13S317 and D7S820--in the Japanese population by multiplex PCR and capillary electrophoresis. J Forensic Sci. 1999;44(1):167-70. https://doi.org/10.1520/jfs14426j
   PMid:9987882
2. Allor C, Einum DD, Scarpetta M. Identification and characterization of variant alleles at CODIS STR loci. J Forensic Sci. 2005;50(5):1128-33. https://doi.org/10.1520/jfs2005024
   PMid:16225220
3. Ellegren H. Microsatellite: Simple sequences with complex evolution. Nat Rev Genet. 2004;5(6):435-45. https://doi.org/10.1038/nrg1348
   PMid:15153996
4. Mats O, Mo H, Erik W, Tonia S, Natasha L, Tobias U. Mating system variation and morph fluctuations in a polymorphic lizard. Mol Ecol. 2007;16(24):5307-15. https://doi.org/10.1111/j.1365-294x.2007.03578.x
5. Mohammed A, Imad H. Autosomal STR: From locus informationto next generation sequencing technology. Res J Biotechnol. 2013;8(10):92-105.
6. Ensenberger MG, Thompson J, Hill B, Homick K, Kearney V, Mayntz-Press KA, *et al*. Developmental validation of the

PowerPlex 16 HS system: An improved 16 locus fluorescent STR multiplex. Forensic Sci Int Genet. 2010;4(4):257-64. https://doi.org/10.1016/j.fsigen.2009.10.007

PMid:20457027

7. Butler JM. Forensic DNA Typing. Biology, Technology and Genetic of STR Markers. 2nd ed. United States: Elsevier Academic Press; 2005. p. 678.
8. Budowle B, Chakraborty R, Carmody G. Source Attribution of a Forensic DNA Profile. Forensic Science Communications; 2000.
9. Central Bureau of Statistic. Number of Population by Gender in Nias Regency. Central Bureau of Statistic; 2020. Available from: https://niaskab.bps.go.id/indicator/12/126/1/jumlah-penduduk-menurut-jenis-kelamin-di-kabupaten-nias-html
10. Van Oven M, Hämmerle JM, Van Schoor M, Kushnick G, Pennekamp P, Zega I, *et al*. Unexpected Island effects at an extreme: Reduced Y chromosome and mitochondrial DNA diversity in Nias. Mol Biol Evol. 2011;28(4):1349-61. https://doi.org/10.1093/molbev/msq300

PMid:21059792.

11. Bar W, Brinkmann B, Budowle B, Carracedo A, Gill P, Lincoln P, *et al*. DNA recommendations-further report of the DNA commission of the ISFH regarding the use of short tandem repeat systems. Int J Legal Med. 1997;110(4):175-6. https://doi.org/10.1007/s004140050061

PMid:9274938

12. Olaisen B, Bar W, Brinkmann B, Budowle B, Carracedo A, Gill P, *et al*. DNA recommendations 1997 of the international society for forensic genetics. Vox Sang. 1998;74(1):61-3. https://doi.org/10.1046/j.1423-0410.1998.7410061.x

PMid:9481867

13. Fung, WK, Yue QH. Statistical DNA Forensics: Theory, Methods and Computation. 1st ed. India: Wiley; 2008. p. 264. Available from: https://saasweb.hku.hk/EasyDNA
14. Imad HH, Aamera JO, Abeer FM, Ghaidaa JM. Allele frequency data of 21 autosomal short tandem repeat loci in mesan and Basra Provinces in South Iraq. Egypt J Forensic Sci. 2015;5(4):150-6. https://doi.org/10.1016/j.ejfs.2014.10.003