List of Publications
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**2004-2005**


**2003-2004**


**2002-2003**


**2001-2002**


**2000-2001**

FOR THE RECORD

Faisal Khan,1,2 M.Sc.; Atul Kr. Pandey,1 M.Sc.; P. S. Bison,2 Ph.D.; and Suraksha Agrawal,1 Ph.D.

Allele frequency profile of three STR loci in nine north Indian populations

Key words: Forensic science, DNA typing, Short tandem repeats, Polymerase chain reaction, Uttar Pradesh-India, D5S818, D7S820 and FGA

Populations: Bhargavas (n= 120), Chaturvedis (n= 120), Brahmins (n= 120) Muslim Sunni (n=120), Muslim Shiya (n=120); Kayastha (n= 120), Mathurs (n= 120), Rastogies (n= 120) and Vaish (n= 120)

Whole blood obtained by venipuncture was collected in EDTA vacutainer tubes from individual residing in different parts of Uttar Pradesh, India. The DNA was extracted by phenol chloroform method (1) and purified by ethanol precipitation. PCR amplification was performed for three autosomal STR loci namely D5S818, D7S820 and FGA using flanking primers (one of the primer for each loci was labeled with fluorescent dye Ned, VIC and 6-FAM respectively) described be Perez-Lezaun et.al, 1997 (2). The amplified products were separated by capillary electrophoresis on ABI 310 genetic fragment analyzer. Genotyping was done with the help of 500-ROX-size standard using GENESCAN v3.4 and GENOTYPER v1 software. The data was analyzed using software POPGENE (3) and CERVUS (4). The allele frequency data is tabulated in Table 1-3 for all the nine populations. The complete data are available to any interested researcher upon request.

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3. POPGENE home page: http://www.ualberta.ca/~fyeh/fyeh


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2. Department of Biotechnology, J.C. Bose Institute of Medical Sciences, Bundelkhand University, Jhansi, India
Reconstructing recent human phylogenies with forensic STR loci: A statistical approach
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Abstract

Background: Forensic Short Tandem Repeat (STR) loci are effective for the purpose of individual identification, and other forensic applications. Most of these markers have high allelic variability and mutation rate because of which they have limited use in the phylogenetic reconstruction. In the present study, we have carried out a meta-analysis to explore the possibility of using only five STR loci (TPOX, FES, vWA, F13A and Tho1) to carry out phylogenetic assessment based on the allele frequency profile of 20 world population and north Indian Hindus analyzed in the present study.

Results: Phylogenetic analysis based on two different approaches – genetic distance and maximum likelihood along with statistical bootstrapping procedure involving 1000 replicates was carried out. The ensuing tree topologies and PC plots were further compared with those obtained in earlier phylogenetic investigations. The compiled database of 21 populations got segregated and finely resolved into three basal clusters with very high bootstrap values corresponding to three geo-ethnic groups of African, Orientals, and Caucasians.

Conclusion: Based on this study we conclude that if appropriate and logistic statistical approaches are followed then even lesser number of forensic STR loci are powerful enough to reconstruct the recent human phylogenies despite of their relatively high mutation rates.

Background

Short Tandem Repeats (STR), with a repetitive sequence ranging from 2–6 base pairs are amongst the most polymorphic markers reported till date. They exhibit substantial allelic variability due to high rate of germline mutations [1]. The STR loci have a uniform and dense distribution throughout the genome and exhibit high level of relatively stable polymorphism [2]. All these features make them an ideal candidate for diverse applications including forensic applications [3], individual identification, paternity/maternity detection [2], fine scale genetic mapping [4] and inter and intra group phylogenetic reconstruction [5].

However, a specific set of STR can be employed for specific applications and this specificity is solely based on the properties of STR loci involved and their suitability to the particular application. STR loci used for forensic purposes
YAP, signature of an African-Middle Eastern migration into northern India

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YAP, an Alu insertion polymorphism found on human Y-chromosomes is present in two lineages worldwide, corresponding to M145/M203/SRY4064 (haplogroup E) and M145/M203/M174 (haplogroup D) polymorphisms respectively. First lineage belonging to haplogroup D is specific to Japan and other Southeast Asian populations, while haplogroup E is confined to Sub-Saharan African, Middle Eastern and Southern European populations. In the present study, 1021 Y-chromosomes belonging to nine different populations of North India were analysed for YAP insertion and four other single nucleotide polymorphisms (SNPs) to delineate the two lineages. Out of nine populations only one, i.e. Shiya Muslims revealed presence of YAP element at a frequency of 11%. Further analysis based on four additional SNPs revealed that all the YAP+ve samples could be categorized under African/Middle East-specific haplogroup E lineage. Interestingly, Sunni Muslims who historically have the same origin, i.e. from the Middle east showed a complete lack of YAP+ve lineage similar to other castes. We hypothesize that unlike Sunnis, Shiya Muslims due to their lesser number and less admixture with other caste groups of India, still carry the ancestral YAP+ve lineage, which in all probabilities is one of the founder haplogroups. All Middle Eastern populations show the presence of this lineage in almost similar frequency. Our study shows the presence of YAP+ve lineage in North Indian populations, reflecting an African/Middle Eastern migration into North India.

The Y-chromosome - a single haploid entity passed from father to son, is a highly suitable marker to trace the patrilineal migration due to its uniparental transmission, lack of recombination and its high sensitivity despite small effective population size. At least three different types of polymorphism have been reported on the Y-chromosome: contraction-expansion mutation at tandem repeats markers, single nucleotide polymorphism (SNP) mutation and insertions of reiterated elements (e.g. YAP polymorphism). The YAP marker originated when an Alu repetitive element retro transposed at the DYS 287 locus at location Yq11. The use of this polymorphic Alu insertion (PAI) in human population studies has been bolstered by DNA sequencing studies that show that the YAP element is inserted be-

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Phylogeny of Mitochondrial DNA Macrohaplogroup N in India, Based on Complete Sequencing: Implications for the Peopling of South Asia

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To resolve the phylogeny of the autochthonous mitochondrial DNA (mtDNA) haplogroups of India and determine the relationship between the Indian and western Eurasian mtDNA pools more precisely, a diverse subset of 75 macrohaplogroup N lineages was chosen for complete sequencing from a collection of >800 control-region sequences sampled across India. We identified five new autochthonous haplogroups (R7, R8, R30, R31, and N5) and fully characterized the autochthonous haplogroups (R5, R6, N1d, U2a, U2b, and U2c) that were previously described only by first hypervariable segment (HVS-I) sequencing and coding-region restriction-fragment-length polymorphism analysis. Our findings demonstrate that the Indian mtDNA pool, even when restricted to macrohaplogroup N, harbors at least as many deepest-branching lineages as the western Eurasian mtDNA pool. Moreover, the distribution of the earliest branches within haplogroups M, N, and R across Eurasia and Oceania provides additional evidence for a three-founder-mtDNA scenario and a single migration route out of Africa.

Introduction

The “population genomics” era has emerged in the research of human mtDNA (Hedges 2000; Richards and Macaulay 2001) by utilization of complete or nearly complete mtDNA sequences to infer the prehistoric dispersal of modern humans and the phylogeny of the major mtDNA lineages in Europe, Africa, America, Oceania (Australia and Papua New Guinea), and East Asia (Ingman et al. 2000; Finnilä et al. 2001; Maca-Meyer et al. 2001, 2003; Torroni et al. 2001; Derbeneva et al. 2002b; Herrnstadt et al. 2002, 2003; Ingman and Gyllensten 2003; Kong et al. 2003; Mishmar et al. 2003; Relea et al. 2003). However, complete phylogenetic information was hitherto not available for South Asia and for India in particular, an area that served as a major corridor of modern human dispersal out of Africa (Cann 2001) and that hosts a diverse conglomerate of people with different morphological, genetic, cultural, and linguistic characteristics. Quite a number of mtDNA studies that focus on the first hypervariable segment (HVS-I) of the control region have been applied to various Indian populations and have provided some insights into the genetic structure of the populations in this area (Kaur et al. 2002; Basu et al. 2003; Kivisild et al. 2003a; Roy et al. 2003 and references therein). In addition to India-specific M subhaplogroups (i.e., M2, M3, M4, M5, and M6), some autochthonous haplogroups, including U2a, U2b, U2c, and many unclassified lineages within the nested macrohaplogroups R and N, have been observed in Indian populations (Passarino et al. 1996; Kivisild et al. 1999a, 1999b, 2003a, 2003b; Bamshad et al. 2001; Basu et al. 2003; Quintana-Murci et al. 2004). However, since these studies were based mainly on HVS-I plus a few coding-region RFLPs, none of those haplogroups had yet been fully characterized. Moreover, some western Eurasian haplogroups also occur in India at low frequencies (Passarino et al. 1996; Kivisild et al. 2003b; Quintana-Murci et al. 2004)—or even at high frequencies, in particular regions (Forster et al. 2002). Although rather detailed phylogenies of western Eurasian mtDNA lineages have been obtained by Finnilä et al. (2001), Maca-Meyer et al. (2001), and Herrnstadt et al. (2002) (which were, however, in minor conflict with one another), no Indian counterpart was hitherto available for comparison. Although most of the mtDNA line-
Review

Significance of chimerism in hematopoietic stem cell transplantation: new variations on an old theme

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Summary:

The main goal of post-transplantation monitoring in hematopoietic stem cell transplantation (HSCT) is to predict negative events, such as disease relapse, graft rejection and graft-versus-host disease, in order to intervene with appropriate therapy. In this context, chimerism analysis is an important method in monitoring post HSCT outcome. Mixed chimerism (MC) is mainly evaluated to define engraftment and relapse. Detection of MC is a prerequisite in both myeloablative and non-myeloablative HSCT, in order to assess the graft status and decide later therapeutic strategies such as donor lymphocyte infusion. In this review, we discuss various techniques including erythrocyte phenotyping, cytogenetic analysis, fluorescent in situ hybridization, restriction fragment length polymorphism, STR/VNTR analysis and real-time quantitative PCR, along with the various methods used to detect minimal residual disease (MRD) in different diseases such as chronic myeloid leukemia, acute myelomonocytic leukemia or acute lymphoblastic leukemia. The review mainly highlights the optimal methodological approach, which needs to be informative, sensitive and quantitatively accurate for MC detection. Future of post HSCT graft monitoring lies in the selection of the most accurate and sensitive technique to determine both MC and MRD. Such an approach would be helpful in not only determining relapse or rejection, but also in ascertaining various responses to different treatment modalities.

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Keywords: chimerism; hematopoietic stem cell transplantation; short tandem repeats; donors' lymphocyte infusion; nonmyeloablative transplant

In Greek mythology, the Chimera was a creature with the head of a lion, the body of a goat and tail of a serpent. In medicine, the term chimera is used to designate an individual whose body contains cell populations derived from different individuals of the same or a different species occurring spontaneously or produced artificially. The phenomenon of co-existence of cells from two different organisms (evolved from two different zygotes) in one body is called chimerism.

In this review, we consider chimerism in allogeneic hematopoietic stem cell transplantation (HSCT), as it is one of the important states that develop after engraftment, and it is an important indication of disease relapse, graft rejection or graft-versus-host disease (GVHD). Over the past two decades, allogeneic HSCT has become the treatment of choice for patients suffering from certain malignant and nonmalignant hematological disorders. Allogeneic HSCT has been effective in the reconstitution of normal hematopoiesis in these patients. Furthermore, allogeneic HSCT is the preferred therapeutic option, primarily because of its intrinsic graft-versus-leukemia (GVL) effect. Most of this GVL effect is usually ascribed to donor T-cell immunoreactivity against host minor histocompatibility antigens, developmentally regulated antigens or leukemia-specific epitopes.

The success of this treatment modality is mainly affected by the recurrence of the underlying disease. Factors responsible for relapse include insufficient conditioning regimens or, eventually, a deficient GVL effect due to decreasing amounts of effector cells or to their functional ineffectiveness. In human HSCT, complete donor-derived hematopoiesis has been considered essential for sustained engraftment and for the prevention of relapse. Successful chemotherapy/radiotherapy causes eradication of all hematopoietic progenitors, which results in stable chimerism. Different states of chimerism are summarized in Table 1. The stage when the patient shows no evidence of recipient cells at any time after transplantation is considered to be complete chimerism (CC). The second stage is mixed chimerism (MC), where the patient shows both recipient as well as donor cells in the peripheral blood. However, the recipient cells in MC could be normal hematopoietic cells or leukemic cells. Persistence of residual leukemic cells is a result of the inefficiency of ablative conditioning regimens. With time, following HSCT, it is possible that these cells will re-emanate, resulting in leukemia relapse. The presence
Genetic variation of ApoB 3’ hyper variable region polymorphism among Brahmins of North India

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ApoB 3’ hyper variable region (ApoB 3’HVR) is highly polymorphic and hence an informative marker. It could be an ideal candidate to study the genetic heterogeneity among different population groups of the Indian subcontinent. It is one of the markers for which population data are available. This makes the ApoB 3’HVR an ideal locus for a pilot study to investigate the relationships between different populations and the micro-evolutionary processes leading to their present-day distribution. In the present investigation, we have studied ApoB 3’HVR in three endogamous groups of North India and have compared these populations on the basis of inter- and intra-group diversity. The sub-populations chosen were Bhargavas, Chaturvedis, and non-Bhargava non-Chaturvedi Brahmins of Uttar Pradesh. Nineteen segregating alleles were detected in our population groups. The average observed heterozygosity was quite high (0.717), suggesting high diversity at the ApoB 3’HVR locus. Low value of average Gst (0.0126) and Fst (0.002) reflects non-significant deviation of heterozygosity between the three subgroups. On comparing the three study groups with ApoB 3’HVR of other Indian and world populations, it was clear that greater diversity was observed for Africans followed by Europeans and Asians. There was relative homogeneity among the Continental groups. In our study it was observed that there was high heterozygosity, an extended range of allele size, a quasi unimodal allele size distribution, centred on HVR 37. These findings indicate that our populations may be characterized as ancestral, since similar features are observed in the African population. ApoB 3’HVR polymorphism suggests that despite practising restricted natal patterns, these groups or castes do not significantly differ from each other at the genetic level. This may be because of the fact that divergence time may not be enough to cause genetic variation in these groups. However it may not be ruled out that the ApoB 3’HVR polymorphism probably predate the divergence of these sub-castes. We are further testing this observation, using mt-DNA for maternal lineages and Y-chromosome markers for paternal lineages.

MINISATELLITES – the tandem repeats of sequences ranging from 10 to 70 base pairs, are amongst the most polymorphic markers reported till date. They exhibit substantial allelic variability in the number of repeat units as a consequence of high rate of germine mutations leading to new allelic states.

Among the numerous minisatellites known so far, the one located about 75 bp downstream from the 3’ end of the apolipoprotein-C coding gene is a hyper variable region, designated as ApoB 3’HVR. This locus is highly polymorphic and until now about 23 alleles have been reported. ApoB 3’HVR consists of an AT-rich core repeat sequence of 15 bp. Two basic types of 15-bp repeats (X and Y) have been identified3,4. Presence of high allelic variability at ApoB 3’HVR is due to the complex mutational pattern. Earlier studies have reported that stepwise mutational model (SMM), which reflects gain or loss of one or few repeat units probably due to replication slippage, is responsible for creating high polymorphism at ApoB 3’HVR.

All these features make ApoB 3’HVR a useful marker for population studies at the genetic level. The availability of numerous population data makes the ApoB 3’HVR an ideal locus for a pilot study to find out the relationships between different populations on the basis of allele frequency distribution and also the micro-evolutionary processes leading to their present-day distribution.

As ApoB 3’HVR, is a highly polymorphic and informative marker, it is ideal to study the genetic heterogeneity among different groups in India. Our previous studies5-6 indicated that the North Indians of Uttar Pradesh (UP) occupy an intermediary zone which has Caucasoid, Negroid, Australoid and Mongoloid elements. Another important feature of Indian populations is caste system, which is the basic element of the Indian social structure7.

The major caste groups in India are Kshatriyas, Brahmins and Vaisyas; they are subdivided into several subgroups (identified by ‘gotras’ and surnames) and marriage within the same gotra is generally not preferred/allowed. In North India, a further degree of endogamy is seen in some sects/groups where marriages are restricted within the same surname. Two such groups are Bhargavas and Chaturvedis. They belong to the broad caste group of Brahmins, but they do not marry outside their own surnames. Thus, Bhargavas marry within Bhargavas and Chaturvedis within Chaturvedis, and not with other Brahmins.

In the present investigation, we have studied ApoB 3’HVR in three endogamous groups of North India and have compared these populations on the basis of within group diversity (in terms of heterozygosity, number of alleles, and allele size distribution), between group diversity (in terms of total genomic diversity and coefficient of gene differentiation) and Wright’s F-statistics (in terms of fixation index). The sub-populations chosen for the study were Bhargavas, Chaturvedis and non-Bhargava non-Chaturvedi Brahmins of UP, where the former two are the subsets of Brahmins. They follow endogamy within same sub-caste and have a well-defined pattern of exogamy within the same gotra. Thus, there is no consanguinity in these subpopulations in spite of surname endogamy.

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Original communication

Use of ApoB3' hyper variable region in studying mixed
chimerism and maternal contamination in North Indian
populations

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Abstract

ApoB3' hyper variable region is one of the highly polymorphic genetic marker and reveals a high degree of allelic variation in
different populations therefore; it can be a useful marker for different clinical tests in which individual differences at DNA level form
the basis of detection. In the present study we compared Apo B3 HVR with other 28 STR markers at allele frequency level, heterozygosity,
polymorphism information content (PIC) and power of exclusion. Our results indicated a high degree of heterozygosity, PIC and power of exclusion for Apo B3 HVR. These criteria lead us to investigate this marker for different purposes like detection of
maternal contamination in chorionic villus samples and chimerism studies after the engraftment of bone marrow in bone marrow
transplantation patients. The utility of this marker has been discussed in comparison of other markers.

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Keywords: ApoB3'HVR; Polymorphism information content; Chimerism; Maternal contamination; Chorionic villus sampling

1. Introduction

Data generated by human genome sequencing project indicate that any two randomly drawn humans are genetically about 99.9% identical. Human genetists all
over are studying human genome diversity based on this tiny fraction of DNA. It is this small fraction that
confers the element of uniqueness to every human. It is primarily this fraction on which various evolutionary
forces have acted during the course of evolution. Differences in this small fraction make some individual
susceptible to a disease, while conferring protection to others from the same disease. This element of uniqueness is employed for various purposes including forensic
applications and other molecular tests, which are of clinical relevance.1

One of the utility is in evaluating chimerism in allogenic BMT, which is an important state which develop after engraftment. On the basis of chimerism we can know about the disease relapse, graft rejection or GVHD. Over past two decades, allogenic BMT has become the treatment of choice for patients suffering from malignant hematological disorders [acute myeloid
leukemia (AML), chronic myeloid leukemia (CML) and acute lymphoblastic leukemia (ALL)], severe cytopenias
[myelodysplastic syndrome (MDS) and severe aplastic anemia (AA)] and other non-malignant hematological
disorders (α- and β-thalassemia, severe combined immunodeficiency, haemoglobinopathies), etc.5-14 Allogenic
BMT has been effective in reconstitution of normal haemopoiesis in these patients. The success of this
treatment modality is mainly affected by the recurrence of the underlying disease. Chimerism studies are the
most sensitive tests used for monitoring the graft status after bone marrow transplantation. These tests are
based on detection of informative polymorphic markers, which are regions of DNA, which differ in-patient and
donor.5,7 These regions could be different restriction fragment length polymorphism (RFLP) sites. STRs,
VNTRs or sex specific markers. This is followed by the study of these loci in patients after transplantation,
which helps in establishment of graft status as complete chimerism (complete engraftment), mixed chimerism
(partial engraftment) or graft failure.
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Evaluation of Four Short Tandem Repeat Loci among North Indian Muslim Populations

POPULATIONS: Muslim Sunni (n = 200), Muslim Shia (n = 200)

KEYWORDS: forensic science, DNA typing, short tandem repeats, polymerase chain reaction, population genetics, Uttar Pradesh-India, TPO, vWA, Th01, vWF-1, FES

Whole blood obtained by venipuncture was collected in EDTA vacutainer tubes from individual residing in different parts of Uttar Pradesh, India. The DNA was extracted by phenol chloroform method (1) and purified by ethanol precipitation. PCR amplification was performed using flanking primers described by Perez-Lezaun et al. (2). The amplified product was separated and detected on 9% PAGE using silver staining. The data were analyzed using software POPGENE (3) and CERVUS (4). The allele frequency data is tabulated in Table 1.

The complete data are available to any interested researcher upon request.

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3. POPGENE home page: http://www.uslberta.ca/~fyeh/fyeh

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Microsatellite Variation at 24 STR Loci in Three Endogamous Groups of Uttar Pradesh, India

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Abstract We have studied variation at 24 microsatellite markers among 50 individuals from each of three endogamous groups, Bhargava, Chaturvedi, and non-Bhargava, non-Chaturvedi Brahmins of Uttar Pradesh, India. The number of alleles at the loci tested varied from 4 to 10, with an average of 6 at each locus. Heterozygosity was found to be quite high at all loci in the three subpopulations. It varied between 0.44 to 0.84 among Bhargavas (average 0.6510±), 0.44 to 0.80 among Chaturvedis (average 0.663±), and 0.42 to 0.85 among Brahmins (average 0.694±). Hardy-Weinberg equilibrium analysis revealed that these populations are under genetic equilibrium at almost all the loci tested. Comparisons of allele frequency between Bhargavas and Chaturvedis showed that they differed significantly at 14 short tandem repeat (STR) markers (p < 0.001), while Chaturvedis and Brahmins differed at 6 (p < 0.05) and Brahmins and Bhargavas at 8 (p < 0.05). Average $F_{ST}$ and $F_{ST}$ for the 24 STR markers was 0.02 and 0.013, respectively. We used both unweighted pair group with arithmetic mean and principal components analysis to evaluate genetic distances among the three groups. Our results revealed that although there were differences at particular allele frequencies between Bhargavas vs. Brahmins, Bhargavas vs. Chaturvedis, and Brahmins vs. Chaturvedis, these differences were not statistically significant when combined over all 24 STR markers between Chaturvedis vs. Brahmins and Bhargavas vs. Brahmins. The genetic distance analysis revealed that Bhargavas are slightly apart from the other two populations.

The caste system, a basic institution of the Indian social structure, is so elaborate and pervasive that no aspect of social life in India remains untouched by it. The multiplicity of caste can be illustrated by the fact that Hutton (1961) has enumerated about 3000 castes in India. Caste has been defined by Karve (1961) as an

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KEY WORDS: STR MARKERS, SURNAME ENDOGAMY, GENETIC DISTANCE; BHARGAVA, CHATURVEDI, BRAHMIN, NORTH INDIA
Evaluation of Six Short Tandem Repeat Loci in Forensics: North Indian Populations

ABSTRACT: In a North Indian population a study was performed on the STR markers D6S1006, D6S1007, D7S2201, D8S592, D11S2371, and D12S1300. Each of the six STR markers were analyzed by using ABI 373A genetic analyzer (Applied Biosystem, Inc.). This study was done on a sample of 150 unrelated North Indians (Uttar Pradesh) from three different caste groups to determine allele frequencies at these STR loci, and to create the database for North Indians from Uttar Pradesh, India.

KEYWORDS: forensic science, DNA, STR, North Indian population, Bhargava, Chaturvedi, Brahmin

Population: Fifty healthy unrelated individuals were randomly chosen from each of the following three populations: Bhargava, Chaturvedi, and Brahmin. Three-generation pedigree charts were prepared to ensure (i) surname endogamy in Bhargavas and Chaturvedis, and caste endogamy in Brahmins. Subjects were chosen from several parts of Uttar Pradesh, a northern state of the Indian republic.

Extraction: Modified salting out technique (1) followed by phenol-chloroform extraction and ethanol precipitation.

PCR
1–2 ng of target DNA, using flanking primers (2–4) purchased from Research Genetics, one of which was fluorescently labeled.

Typing: ABI 373A genetic analyzer, Genescan T, Genotype T.
Analysis of data: POPGENE (5) TFPGA (6) CERVUS (7)

Results
See Tables 1–3.

Other Remarks
The allele frequency estimates of these STR markers reveal that alleles are not equally distributed in all three populations included in the study.

Alleles at all the loci were in Hardy Weinberg equilibrium. There was no nonrandom association between alleles at two different loci. Markers are informative and can be used for forensic DNA analysis and paternity testing.

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5. POPGENE home page: http://www.ualberta.ca/~fye/fye

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FOR THE RECORD

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Distribution of Allele Frequencies of Six STR Markers in North Indians

POPULATION: Fifty healthy unrelated individuals were randomly chosen from each of the following three populations viz., Bhargava, Chaturvedi, and Brahmin. Three-generation pedigree charts were prepared to ensure (1) surname endogamy in Bhargavas and Chaturvedies, and caste endogamy in Brahmins. Subjects were chosen from several parts of Uttar Pradesh, a northern state of the Indian republic.

KEYWORDS: forensic science, DNA, STR, North Indian population, Bhargava, Chaturvedi, Brahmin

Extraction: Modified salting out technique (1) followed by phenol—chloroform extraction and ethanol precipitation.

PCR

1–2 ng of target DNA, using flanking primers (2–4) purchased from Research Genetics, one of which was fluorescently labeled.

Typing: ABI 373A genetic analyzer, Genescan T, Genotype T.


Results

See Tables 1–3.

Other Remarks

The allele frequency estimates of these STR markers reveal that alleles are not equally distributed in all of the three populations included in the study.

Alleles at all the loci were in Hardy Weinberg equilibrium. There was no non-random association between alleles at two different loci. Markers are informative and can be used for forensic DNA analysis and paternity testing.

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5. POPGENE home page: http://www.alberta.ca/~fyeh/fyeh

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FOR THE RECORD

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Allele Frequencies of Microsatellite Repeat Loci in Bhargavas, Chaturvedis, and Brahmins of North India

POPULATION: Fifty healthy unrelated individuals were randomly chosen from each of the following three populations viz., Bhargava, Chaturvedi, and Brahmin. Three generation pedigree charts were prepared to ensure (i) surname endogamy in Bhargavas and Chaturvedies, and caste endogamy in Brahmins. Subjects were chosen from several parts of Uttar Pradesh, a northern state of the Indian republic.

KEYWORDS: forensic science, DNA, STR, North Indian population, Bhargava, Chaturvedi, Brahmin

Extraction: Modified salting out technique (1) followed by phenol—chloroform extraction and ethanol precipitation.

PCR
1–2 ng of target DNA, using flanking primers (2–4) purchased from Research Genetics, one of which was fluorescently labeled. Typing: ABI 373A genetic analyzer, Genescan T, Genotype T. Analysis of data: POPGENE (5) TFPGA (6) CERVUS (7)

Results
See Tables 1 to 3.

Other Remarks
The allele frequency estimates of these STR markers reveal that alleles are not equally distributed in all three populations included in the study. Alleles at all the loci were in Hardy Weinberg equilibrium. There was no nonrandom association between alleles at two different loci. Markers are informative and can be used for forensic DNA analysis and paternity testing.

References
5. POPGENE home page: http://www.uaalberta.ca/~fyeh/fyeh
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DNA Short Tandem Repeat Profiling of Three North Indian Populations

Population: Fifty healthy unrelated individuals were randomly chosen from each of the three populations viz., Bhargavas, Chaturvedies, and Brahmins. Three generation pedigree charts were prepared to ensure surname endogamy in Bhargavas Chaturvedies and group endogamy in Brahmins subjects were chosen from several parts of Uttar Pradesh, a northern state of the Indian republic.

KEYWORDS: forensic science, DNA, STR, North Indian populations, Bhargavas, Chaturvedies, Brahmins.

In a North Indian population, a study was performed on the STR markers D20S115, D1S1728, D2S1329, D6S1270, D12S398, and D15S816. Each of the six STR markers were analyzed by using ABI 373 A genetic analyzer (Applied Biosystem, Inc.). This study was done on a population of 150 unrelated North Indians (Uttar Pradesh) from three different caste groups to determine allelic frequencies at these STR loci and to create the database for North Indians from Uttar Pradesh, India.

Results—See Table 1 to 3.

Access to Data—Via electronic mail from corresponding author at suraksha@sgpgi.ac.in

Other Remarks

The allele frequency estimates of these STR markers reveal that alleles are not equally distributed in all the populations. All follow Hardy Weinberg equilibrium. These were no nonrandom association between alleles at two different loci. Markers are informative and can be used for forensic DNA analysis and paternity testing.

References

5. POPGEO home page: http://www.slab.ca/~fyeh/fyeh

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