

Exploring Population Diversity in India through Genetic Studies: A Review

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ABSTRACT

Because of migrations, endogamy, and sociocultural relations India is rich in its genetic diversity. This review highlighted findings regarding genetic diversity from available genetic studies utilizing Short Tandem Repeat (STR) markers, in India's population and relation of genetic diversity with forensic implications. Studies on populations like the Tamil, Bhil, Gorkha, and Malayalam-speaking communities reveal unique allelic distributions and genetic affinities. Forensic parameters, including Polymorphic Information Content (PIC) and Power of Discrimination (PD), underscore the utility of STR markers in criminal investigations and paternity testing. Population comparison studies elucidate genetic relatedness, with distinct clusters observed based on geographical and linguistic affiliations. For instance, the genetic distance analysis of the Tamil population compared to global and regional populations highlights unique genetic affinities and substructures. Despite advancements, noticeable gaps remain, particularly in northwestern India, urging comprehensive research to enhance forensic DNA analysis and ensure justice in legal proceedings. The absence of allele frequency data specific to regions like Chhattisgarh underscores the need for targeted studies to improve DNA profiling accuracy. The study will help in exploring India's genetic complexities, information on historical migrations and creation of a database for advancing forensic science for the benefit of the criminal justice system.

Keywords: Genetic diversity, Short Tandem Repeat markers, Indian populations, Forensic genetics, Population comparisons.

 https://doi.org/10.5281/zenodo.13934540

BACKGROUND

India, with its vast and diverse population, boasts a unique and intricate genetic landscape. Hosting over a billion people, the Indian subcontinent includes a multitude of ethnic, linguistic, cultural, and religious groups, each adding to the complexity of its genetic diversity. The region's extensive history of migrations, invasions, and settlements has further contributed to this rich genetic mosaic, making it a significant focus for genetic and anthropological research.

Investigating population diversity in India through genetic analysis provides vital insights into human migration patterns, evolutionary history, and adaptation mechanisms. These genetic studies are crucial not only for understanding human history but also for advancing medical research, public health, and forensic science. By analyzing genetic variations and commonalities among different groups, researchers can identify disease susceptibility patterns, develop personalized medical treatments, and refine forensic techniques.

Recent advancements in genetic technologies, such as high-throughput DNA sequencing and bioinformatics, have dramatically enhanced our ability to study genetic diversity with unprecedented accuracy and scale. These technological innovations have facilitated the examination of complex genetic traits and the identification of genetic markers linked to various diseases and conditions prevalent in the Indian population.

This review aims to provide a thorough overview of the current state of genetic studies on population diversity in India. It will explore the historical context, spotlighting key research milestones and pioneering contributions. Additionally, it will examine the methodologies used in these studies, the challenges encountered, and the potential applications of the findings in fields such as medicine, anthropology, and forensic science. Through this review, we aim to highlight the significance of genetic research in deciphering India's intricate genetic landscape and its broader implications for science and society.

 https://doi.org/10.5281/zenodo.13934540

MAIN TEXT:

Introduction:

India, with its rich tapestry of cultures, languages, and traditions, serves as a captivating microcosm of human diversity. Spanning a vast expanse, the subcontinent is home to thousands of distinct population groups, each bearing a unique genetic legacy shaped by centuries of historical, social, and geographical factors. The current state of forensic science encompasses a wide range of techniques used to solve crimes, spanning personal, professional, and financial domains. Among the various investigative aspects of forensic science, one of the most critical is the ability to individualize biological evidence relevant to a case. DNA serves as a pivotal form of evidence for establishing an individual's identity. However, as DNA evidence is classified as pattern evidence, it is essential to establish similarities between the alleles present in both the test and reference samples. Therefore, understanding the typical distribution of each allele within a population becomes crucial. Over the past few decades, there has been a notable increase in the utilization of genetic material, particularly DNA typing methods, for individualization purposes. Beginning in the 1970s, advancements in DNA technology paved the way for the exploration of variations or polymorphisms in specific DNA sequences, shifting the focus of polymorphism analysis from DNA output, such as proteins, to DNA itself. In the forensic context, DNA analysis is subsequently divided into five subcategories: RFLP, AmpFLPS, STR, allele-specific oligonucleotides (ASO), and mt-DNA.

Within this diverse landscape, genetic studies have emerged as powerful tools, employing Short Tandem Repeat (STR) markers to unravel the intricate tapestry of India's population diversity. These studies offer not only valuable insights into population structure and migration patterns but also shed light on the forensic characteristics of various Indian communities.

Over the years, genetic research in India has yielded significant findings, enriching our understanding of the genetic makeup of different population groups. However, despite these advancements, noticeable gaps in research coverage persist, particularly in regions such as northwestern India. This review aims to synthesize the findings from diverse genetic studies conducted across India, with the goal of illuminating the genetic landscape and emphasizing the need for comprehensive investigations to bridge existing gaps.

https://doi.org/10.5281/zenodo.13934540

India's genetic landscape mirrors its cultural tapestry, showcasing millennia of migrations, interactions, and adaptations. From the towering peaks of the Himalayas to the sun-drenched shores of the Indian Ocean, the subcontinent harbors a mosaic of genetic diversity. Genetic studies utilizing autosomal STR markers serve as windows into this complexity, unveiling the intricate interplay of ancestral migrations, endogamy practices, and social structures.

Research focused on specific population groups, such as the Tamil, Bhil tribal, Gorkha, and Malayalam-speaking communities, has provided invaluable insights into allele frequency distributions, heterozygosity levels, and deviations from Hardy-Weinberg Equilibrium (HWE). These studies underscore the multifaceted nature of India's genetic heritage, characterized by a myriad of genetic signatures shaped by historical events and cultural practices.

The Tamil population, for instance, has been a subject of intense genetic scrutiny, with studies comparing its allelic data to global and regional populations. Through Nei's genetic distance, Neighbor-Joining Tree, PCA plot, and AMOVA analyses, researchers have delineated distinct clustering patterns, highlighting the genetic affinity of Indian populations while also revealing unique genetic contributions from the Middle East, Africa, and Pakistan.

Similarly, investigations into tribal communities like the Bhil and Gorkha populations have uncovered fascinating genetic affinities and divergence patterns. Through NJ tree and PCA analyses, researchers have elucidated the genetic connections between these tribal groups and their geographical neighbors, shedding light on historical migration routes and interactions.

Furthermore, studies on linguistic communities, such as the Malayalam-speaking population, have provided valuable insights into the genetic relatedness with central and North Indian populations. This genetic affinity, possibly influenced by historical migrations, underscores the complex interplay between linguistic, cultural, and genetic factors in shaping India's population diversity.

Forensic utility lies at the heart of genetic research in India, with STR markers serving as indispensable tools in criminal investigations, paternity testing, and human identification. Through the evaluation of parameters like Polymorphic Information Content (PIC), Power of Discrimination (PD), and Probability of Exclusion (PE), researchers have demonstrated the efficacy of these markers in forensic applications across diverse Indian populations.

 https://doi.org/10.5281/zenodo.13934540

Population comparison studies have further enriched our understanding of India's genetic landscape, unveiling patterns of genetic relatedness and affinity among different population groups. Through analysis of genetic distances and clustering patterns, researchers have elucidated both intra- and inter-group variations, shedding light on historical migrations, admixture events, and sociocultural interactions.

Notably, comparisons between caste and tribal populations have revealed distinct genetic signatures, reflecting both historical and social factors. While some groups exhibit close genetic affinities, others demonstrate significant genetic differentiation, underscoring the complex interplay of genetic drift, gene flow, and social structure.

Despite the wealth of studies on genetic diversity and forensic utility of STR markers in Indian populations, significant gaps remain. The northwestern region of India, in particular, has been relatively understudied, highlighting the need for comprehensive population genetic studies in this area. Additionally, while DNA technology has been employed in forensic casework in Rajasthan since 2011, there is a lack of allele frequency data specific to the Rajasthan population, essential for accurate statistical interpretation of DNA evidence in legal proceedings.

Forensic Utility of Genetic Markers:

The forensic utility of genetic markers has revolutionized the field of forensic science, offering invaluable tools for the identification and resolution of criminal cases. Over the past several decades, advancements in genetic marker technology have significantly enhanced the ability to individualize biological evidence, providing crucial support to investigative efforts worldwide. By exploiting the unique genetic signatures present in an individual's DNA, forensic scientists can establish links between suspects, victims, and crime scenes with unprecedented precision and reliability.Since the emergence of DNA technology in the 1970s, forensic scientists have continually refined and expanded their repertoire of genetic markers, harnessing the power of these molecular tools to unravel complex criminal mysteries. The evolution of DNA typing methods, from restriction fragment length polymorphism (RFLP) analysis to short tandem repeat (STR) profiling and beyond, has not only accelerated the pace of forensic investigations but has also widened the scope of cases amenable to genetic analysis.

In the realm of forensic science, the utilization of genetic markers, particularly Short Tandem Repeat (STR) markers, has emerged as a cornerstone in various investigative processes. These genetic markers, characterized by their polymorphic nature and high discriminatory power, offer invaluable insights into population diversity while simultaneously serving as potent tools in forensic investigations. Parameters such as Polymorphic Information Content (PIC), Power of Discrimination (PD), and Probability of Exclusion (PE) stand as crucial metrics in evaluating the forensic utility of autosomal and Y chromosomal STR markers across the diverse spectrum of Indian populations.

Studies conducted in this domain have consistently demonstrated the efficacy of these genetic markers in forensic applications, spanning criminal investigations, paternity testing, and human identification. By leveraging the unique genetic signatures embedded within these markers, forensic analysts can accurately reconstruct genetic profiles, thereby aiding in the identification of individuals and the resolution of legal cases. Moreover, the comprehensive assessment of forensic parameters has not only underscored the practical significance of these markers but has also deepened our understanding of the genetic distinctiveness prevalent among various Indian communities. Through meticulous evaluation, researchers have gained insights into the intricate genetic landscapes, enabling more precise DNA profiling and interpretation in forensic contexts.

Furthermore, population comparison studies have played a pivotal role in enriching our understanding of India's genetic mosaic, unraveling patterns of genetic relatedness and affinity across different population groups. By analyzing genetic distances and clustering patterns, researchers have unearthed both intra- and inter-group variations, shedding light on historical migrations, admixture events, and sociocultural interactions that have shaped India's genetic diversity over time. Particularly noteworthy are the comparisons made between caste and tribal populations, revealing distinct genetic signatures that not only reflect historical lineage but also underscore the profound impact of social dynamics on genetic structure. These findings emphasize the nuanced interplay between genetic drift, gene flow, and social structure, urging researchers to consider both genetic and sociocultural factors in their population studies for a more holistic understanding of India's genetic landscape.

 https://doi.org/10.5281/zenodo.13934540

Genetic Diversity and Forensic Utility of Short Tandem Repeat Markers in Indian Populations

India, renowned for its vast population diversity, harbors numerous genetically distinct castes, tribes, and ethnic groups, contributing to its rich cultural tapestry. With approximately 4693 different population groups, including 2205 communities and 589 segments, India stands as a testament to human diversity **(Singh *et al.*, 2006; Reich *et al.*, 2009).** This immense diversity is not merely a product of geographical variation but also reflects the deep-rooted social structures shaped by centuries of migration and endogamy **(Singh *et al.*, 2006)**. The intricate mosaic of Indian populations presents an ideal landscape for studying genetic diversity and forensic applications of Short Tandem Repeat (STR) markers.

Allelic diversity studies have provided valuable insights into the genetic makeup of various Indian populations. **Balamurugan *et al.* (2010)** investigated allelic diversity in the Tamil population of South India using 15 autosomal STR loci, revealing a wide range of allele frequencies and heterozygosity levels. Similarly, **Ghosh *et al.* (2011)** explored 11 Indian populations belonging to different linguistic groups, showcasing substantial allelic diversity across caste and tribal communities. The genetic diversity among populations such as the Khatri, Baniya, and Jatt Sikh populations of Punjab, as studied by **Giroti and Talwar (2013)**, further underscores the complexity of genetic variation within India. **Ruqaiya Hussain and Mohammad Afzal (2014)** conducted a study on variations at the TPOX STR locus in the North Indian Muslim population. They investigated both observed and expected heterozygosity, along with several forensic parameters, for this locus. Their analysis included constructing a dendrogram, which revealed minimal genetic distance between the Pathan and Ansari population groups. **Chaudhari and Dahiya (2014)** examined the genetic makeup of a Bhil tribal population from Gujarat, India, analyzing 15 STR loci. They found that the Indo-European Bhil tribe of South Indian Gujarat shares genetic affinities with Dravidian groups. All loci, except vWA and D18S51, were found to be in Hardy-Weinberg equilibrium. **Manpreet Kaur and Badaruddoza (2014)** investigated genetic polymorphism using five STR markers across four Punjabi population groups in India. Their study revealed the highest observed heterozygosity among Ramdasia populations, compared to Jat Sikh, Majbi Sikh, and Brahmins. Additionally, studies on Bhil tribes **(Shrivastava *et al.*, 2016)**, Gorkha populations **(Preet *et al.*, 2016)**, and Malayalam-

 https://doi.org/10.5281/zenodo.13934540

speaking communities **(Sreekumar *et al.*, 2020)** have elucidated unique allelic frequencies and heterozygosity patterns, reflecting the diverse genetic landscape of India.

Forensic interest parameters provide crucial metrics for assessing the utility of STR markers in forensic applications. Studies have evaluated parameters such as Polymorphic Information Content (PIC), Power of Discrimination (PD), Probability of Exclusion (PE), Matching Probability (PM), and Paternity Index (PI) across various Indian populations. **Balamurugan *et al.* (2010)** demonstrated high PIC values and discriminatory power in the Tamil population, highlighting the forensic relevance of STR markers. Similarly, studies on populations like Banias, Khatris, and Jat Sikhs **(Giroti *et al.*, 2013)**, Bhil **tribes (Shrivastava *et al.*, 2016), Gorkhas (Preet *et al.*, 2016)**, and Malayalam-speaking communities **(Sreekumar *et al.*, 2020)** have shown significant forensic efficacy, with high PD and PE values ensuring reliable individual identification and paternity testing.

Shrivastava et al. (2019) assessed the forensic characteristics of loci present in the Powerplex 21 autosomal and Powerplex 23 Y-STR kits using 168 unrelated individuals from Uttar Pradesh, India. Their findings revealed that Penta E exhibited the highest discrimination power (0.980), while CSF1PO showed the lowest (0.855) within the studied population. This data underscores the high polymorphism and utility of the STR multiplex system in forensic inquiries.

Population comparison studies have elucidated the genetic relatedness and differentiation among Indian populations. Nei's genetic distance analysis and population clustering techniques have revealed distinct genetic clusters based on geographical proximity, linguistic affiliations, and historical migrations. For instance, **Balamurugan *et al.* (2010)** compared Tamil populations with global and regional populations, highlighting unique genetic affinities and population substructures. **Ghosh *et al.* (2011)** observed significant genetic differentiation among caste and tribal populations, with Tibeto-Burman populations showing greater genetic distances from Indo-European groups. Similarly, studies on Banias, Khatris, and Jat Sikhs **(Giroti *et al.*, 2013)** , Bhil tribes **(Shrivastava *et al.*, 2016)** , Gorkhas **(Preet *et al.*, 2016)** , and Malayalam-speaking populations **(Sreekumar *et al.*, 2020)** have revealed genetic affinities with specific regional or linguistic groups, reflecting historical migrations and social dynamics.

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Despite the wealth of studies on genetic diversity and forensic utility of STR markers in Indian populations, significant gaps remain. The northwestern region of India, in particular, has been relatively understudied, highlighting the need for comprehensive population genetic studies in this area. Additionally, while DNA technology has been employed in forensic casework in Rajasthan since 2011, there is a lack of allele frequency data specific to the Rajasthan population, essential for accurate statistical interpretation of DNA evidence in legal proceedings. Moreover, understanding the distribution of alleles on different STR markers used in forensic DNA analysis is imperative for improving the accuracy and reliability of DNA profiling in Rajasthan.

In conclusion, the diverse landscape of Indian populations offers a unique opportunity to explore genetic variation and its forensic implications using STR markers. By addressing existing gaps in research and expanding studies to underrepresented regions such as northwestern India, researchers can contribute to a more comprehensive understanding of India's genetic diversity. Moreover, generating population-specific allele frequency data, particularly in states like Rajasthan, is crucial for enhancing the effectiveness of forensic DNA analysis and ensuring justice in legal proceedings. Ultimately, continued research in this field holds the promise of not only unraveling the genetic complexities of India but also advancing forensic science for the benefit of society as a whole.

Comparison Study on Population Demographics within Indian Communities.

The research conducted by **Balamurugan *et al.* (2010)** compared the allelic data of the Tamil population with global and regional populations using Nei's genetic distance, Neighbor-Joining Tree, PCA plot, and AMOVA. They observed distinct clustering patterns, with Indian populations forming one cluster and Middle Eastern and African populations forming another. Pakistan's population formed a separate cluster, closer to the Indian cluster. **Ghosh *et al.* (2011)** analyzed genetic distances among eleven Indian populations, noting the largest genetic distance between caste and tribal populations. They found Tibeto-Burman populations showing greater genetic distances from Indo-European groups, while Brahmin populations from different geographical regions and linguistic groups exhibited the least genetic differentiation, indicating their genetic relatedness. **Giroti *et al.* (2013)** conducted an AMOVA test, revealing significant genetic differences between Banias and Khatris, although

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not among Banias, Khatris, and Jat Sikh populations. They concluded that the studied loci were highly effective for forensic and anthropological studies. **Shrivastava *et al.* (2016)** observed a closer genetic affinity of Bhils with Gond populations, clustering with tribal rather than higher caste populations. They also found the Gorkha population showing genetic affinity with populations from Nepal and Tibeto-Burman groups. **Shrivastava *et al.* (2017)** noted genetic distinction between caste and tribal populations, with Brahmin populations exhibiting genetic closeness to previously reported Brahmin populations. **Imam *et al.* (2017)** compared the population of Jharkhand with previously reported Indian populations, observing consistent results between NJ and PCA analyses. **Sreekumar *et al.* (2020)** investigated the genetic relatedness of the Malayalam-speaking population of Kerala with previously reported populations, suggesting a genetic affinity with central and North Indian populations, possibly due to historical migration.

CONCLUSION

The synthesis of genetic studies on Indian populations utilizing Short Tandem Repeat (STR) markers reveals a complex tapestry of genetic diversity and forensic utility. Through comprehensive investigations, researchers have illuminated the intricate interplay of historical migrations, sociocultural dynamics, and genetic adaptation shaping India's genetic landscape.

Studies focusing on specific population groups such as the Tamil, Bhil tribal, Gorkha, and Malayalam-speaking communities have uncovered unique allelic distributions and genetic affinities, underscoring the multifaceted nature of India's genetic heritage. These findings, elucidated through various genetic distance analyses and population clustering techniques, highlight both intra- and inter-group variations reflective of historical interactions and demographic processes.

Furthermore, the forensic implications of genetic research using STR markers are profound, with parameters like Polymorphic Information Content (PIC), Power of Discrimination (PD), and Probability of Exclusion (PE) demonstrating the efficacy of these markers in criminal investigations, paternity testing, and human identification. Population comparison studies have further enriched our understanding, revealing distinct genetic signatures between caste

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and tribal populations, emphasizing the importance of considering both genetic and sociocultural factors in forensic applications.

Despite significant advancements, notable research gaps persist, particularly in regions like northwestern India. Comprehensive population genetic studies in these areas are essential to fill these lacunae and provide a more holistic understanding of India's genetic diversity. Additionally, the lack of specific allele frequency data, such as in Rajasthan, poses challenges for accurate forensic DNA analysis, highlighting the need for targeted research efforts.

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