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REVIEW ARTICLE

Gotra exogamy and consanguinity: a genetic and public health view

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ABSTRACT

Consanguineous marriages, defined as unions between biologically related individuals, are associated with an increased risk of autosomal recessive genetic disorders because relatives are more likely to share deleterious alleles inherited from a common ancestor. In contrast, Gotra exogamy in Hindu societies is a socio-cultural practice that discourages marriage within the same paternal lineage, whereas endogamy refers to marriage within a defined social or population group. Although Gotra exogamy may have historically limited certain forms of relatedness, its genetic implications remain indirect and culturally interpreted rather than scientifically established. Population genetic studies in India indicate that consanguinity and long-standing endogamy contribute to an increased prevalence of inherited disorders, estimated at 25-60 per 1,000 births in some communities, depending on regional, socioeconomic, and cultural factors. These risks are primarily attributable to autosomal recessive inheritance resulting from increased carrier overlap among related individuals.

This narrative review examines the relationships among Gotra exogamy, consanguinity, endogamy, and genetic disease risk from sociocultural, genetic, and public health perspectives through a structured review of published literature. The review highlights the importance of culturally sensitive genetic counseling, carrier screening, and community awareness programs in identifying at-risk families and reducing the burden of inherited disorders while respecting cultural traditions and promoting scientific literacy.

Keywords: Consanguineous Marriages, Genetic Counseling, Gotra

Introduction

The Gotra system and scientific perspectives

With advances in modern genetics and population health research, there is growing interest in re-examining traditional social systems through a scientific lens. In India, the Gotra system continues to influence marital practices and is often discussed in relation to genetic inheritance, consanguinity, and inherited disease risk. However, the relationship between cultural lineage systems and genetic relatedness is complex and requires careful scientific interpretation (1–3).

Traditionally, Gotra refers to a lineage or clan in Hindu society that traces descent through the paternal line, often to an ancient sage (Rishi) (4,5). It represents a socio-cultural classification system rather than a biological measure of relatedness. Although individuals belonging to the same Gotra are considered part of a shared ancestral lineage, they may not necessarily be closely related genetically, as Gotra affiliation may also reflect historical, social, or symbolic associations rather than documented biological ancestry (2,6).

For clarity, several related concepts should be distinguished. Gotra exogamy refers to the cultural practice of avoiding marriage within the same Gotra. Exogamy more broadly denotes marriage outside a defined social group, whereas endogamy refers to marriage within a particular caste, community, tribe, or population group. Consanguinity, in contrast, refers to unions between biologically related individuals who share one or more recent common ancestors. From a genetic perspective, consanguinity increases the coefficient of inbreeding (F), which measures the probability that two copies of a gene are identical by descent. Increased inbreeding may lead to greater homozygosity and a higher likelihood of expression of autosomal recessive disorders when both parents carry the same pathogenic variant (7,8).

Among Hindu Brahmins, Gotras are traditionally associated with ancient Rishis, including the Saptarishis and other revered figures such as Bharadwaja. Commonly

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recognized root Gotras include Angirasa, Atri, Gautama, Kashyapa, Bhrigu, Vasistha, Kutsa, and Bharadwaja (4,5). These classifications have historically influenced marital norms, particularly the practice of Gotra exogamy (2,6).

The relationship between Gotra exogamy, consanguinity, and genetic risk has attracted attention in both cultural and scientific discussions. While Gotra exogamy may historically have discouraged certain forms of close-kin marriage in some communities, it should not be regarded as a direct genetic screening system or a reliable marker of biological relatedness. Genetic inheritance is considerably more complex than paternal lineage alone. Although the Gotra system has sometimes been loosely compared with paternal lineage markers such as the Y chromosome, inheritance reflects contributions from both parents across the entire genome (9). Consequently, genetic risk depends primarily on biological relatedness, shared ancestry, population structure, and patterns of consanguinity rather than on cultural lineage labels alone (7,8).

Population genetic studies from India demonstrate that long-standing patterns of endogamy and, in some communities, consanguinity have contributed to distinctive genetic structures and an increased prevalence of certain inherited disorders (8). These effects are largely attributable to increased homozygosity and the greater likelihood that both partners carry the same recessive disease-causing variant. Such risks are more appropriately evaluated through measures of biological relatedness and population genetic history rather than cultural classifications alone (7,8).

Regional diversity further illustrates the complexity of lineage systems in India. Although most Indian communities traditionally follow patrilineal descent, certain populations in South India have historically practiced matrilineal inheritance systems, demonstrating that social concepts of lineage and kinship vary considerably across the country (10,11).

This narrative review examines the intersections among Gotra exogamy, consanguinity, endogamy, population genetics, and public health in India. The objective is not to endorse or challenge any cultural tradition but to evaluate available scientific evidence regarding genetic risk, inheritance patterns, and population structure while maintaining cultural sensitivity. By distinguishing socio-cultural lineage systems from biological measures of relatedness, this review aims to provide a balanced framework for understanding inherited disease risk and the role of genetic counseling, carrier screening, and public health awareness in diverse populations.

Methods

This article was conducted as a narrative review to examine the relationships among Gotra exogamy, consanguinity, endogamy, population genetics, and public health implications in India. A structured literature search was performed using PubMed, Scopus, and Google Scholar to identify relevant studies published in English up to July 2025.

The search strategy included combinations of keywords and Medical Subject Headings (MeSH), where applicable, including “Gotra,” “Gotra exogamy,” “consanguinity,” “consanguineous marriage,” “endogamy,” “population genetics,” “autosomal recessive disorders,” “genetic counseling,” “inbreeding,” “homozygosity,” “genetic disease burden,” and “India.” Boolean operators (AND, OR) were used to refine the search strategy.

Inclusion criteria comprised peer-reviewed original research articles, review articles, population genetic studies, epidemiological investigations, public health reports, and authoritative academic publications addressing socio-cultural marriage practices, genetic relatedness, inherited disorders, and population structure in India.

Exclusion criteria included studies focused exclusively on non-human populations, publications not relevant to the Indian context, duplicate records, conference abstracts lacking sufficient information, and non-English publications.

Method of synthesis

Titles and abstracts of 180 records were screened for relevance, followed by full-text assessment of 60 articles. Additional studies were identified through citation tracking of key publications. Ultimately, 40 articles were included in the qualitative synthesis. The selected literature was reviewed and synthesized to integrate evidence from genetics, epidemiology, anthropology, sociology, and public health. Particular emphasis was placed on distinguishing Gotra as a socio-cultural lineage system, consanguinity as a measure of biological relatedness, and endogamy as a population-level genetic phenomenon.

Genetic Inheritance and Lineage: Relevance to Consanguinity and Population Genetics

Human genetic inheritance involves the transmission of genetic material from both parents through 23 pairs of chromosomes. While cultural lineage systems such as Gotra trace ancestry through the paternal line, biological inheritance reflects contributions from the entire genome and follows multiple inheritance patterns, including autosomal, X-linked, Y-linked, and mitochondrial transmission (12–15).

The Y chromosome is inherited from father to son and is therefore used in population genetics to study paternal ancestry and human migration patterns. Mitochondrial DNA, in contrast, is inherited through the maternal line and provides information about maternal ancestry. Most inherited traits and genetic disorders, however, are determined by autosomal chromosomes inherited from both parents, making overall genetic relatedness considerably more complex than paternal or maternal lineage alone (13–15).

Because Gotra follows a patrilineal lineage framework, it has occasionally been compared with Y-chromosome inheritance. However, genetic relatedness is determined by the entire genome rather than paternal lineage alone. Consequently, assessment of inherited disease risk is more appropriately based on biological relatedness,

family history, carrier status, and population genetic factors (16,17).

Advances in genomics, including whole-genome sequencing, haplotype analysis, and population genetic studies, have further demonstrated the complex and multidimensional nature of human ancestry. These approaches provide a more comprehensive understanding of genetic variation and inherited disease risk than can be inferred from patrilineal lineage systems alone, highlighting the importance of distinguishing socio-cultural traditions from biological mechanisms of inheritance (14–20).

Biological Relatedness, Endogamy, and Recessive Disease Risk

Genetic risk in human populations is influenced by both consanguinity and endogamy, although these processes operate through different mechanisms. Consanguinity refers to unions between biologically related individuals who share recent common ancestry (Figure 1), whereas endogamy refers to marriage within a defined social, caste, tribal, religious, or geographic population over multiple generations. While consanguinity increases genetic relatedness at the family level, long-term endogamy can shape genetic variation at the population level by restricting gene flow and promoting founder effects (8,21).

In the Indian context, the distinction between these concepts is particularly important. Increased prevalence of autosomal recessive disorders may occur not only in populations practicing consanguineous marriage but also in populations with a long history of endogamy, even when marriages are not between close relatives. Repeated marriage within relatively isolated groups can increase the frequency of specific disease-causing variants inherited from a limited number of ancestors, a phenomenon known as the founder effect (22,23).

Consanguinity and endogamy both contribute to increased homozygosity through different pathways. Consanguinity increases the probability that offspring inherit identical alleles from a recent common ancestor, whereas endogamy may increase the frequency of particular pathogenic variants within a population over multiple generations. Consequently, the burden of recessive disorders in India reflects a combination of recent biological relatedness, historical population structure, and founder effects rather than any single cultural practice or lineage classification (22,23). These marriage practices are influenced by diverse social, economic, and cultural factors, including family structure, inheritance patterns, social trust, kinship continuity, family compatibility, and community traditions, which vary across regions and populations (8,21).

From a genetic perspective, consanguinity increases the probability that both partners carry the same inherited genetic variants because of shared ancestry. This is particularly relevant for autosomal recessive disorders, in which disease occurs when an individual inherits two copies of a pathogenic variant, one from each parent. Consanguineous unions therefore increase the likelihood of homozygosity and expression of recessive genetic conditions (8,21) (Figure 2). Such marriages remain culturally prevalent in many parts of the world, accounting for nearly 20% of global marriages.

The increased genetic risk associated with consanguinity can be quantified using the coefficient of inbreeding (F), which estimates the probability that two alleles are identical by descent. For first-cousin unions, F

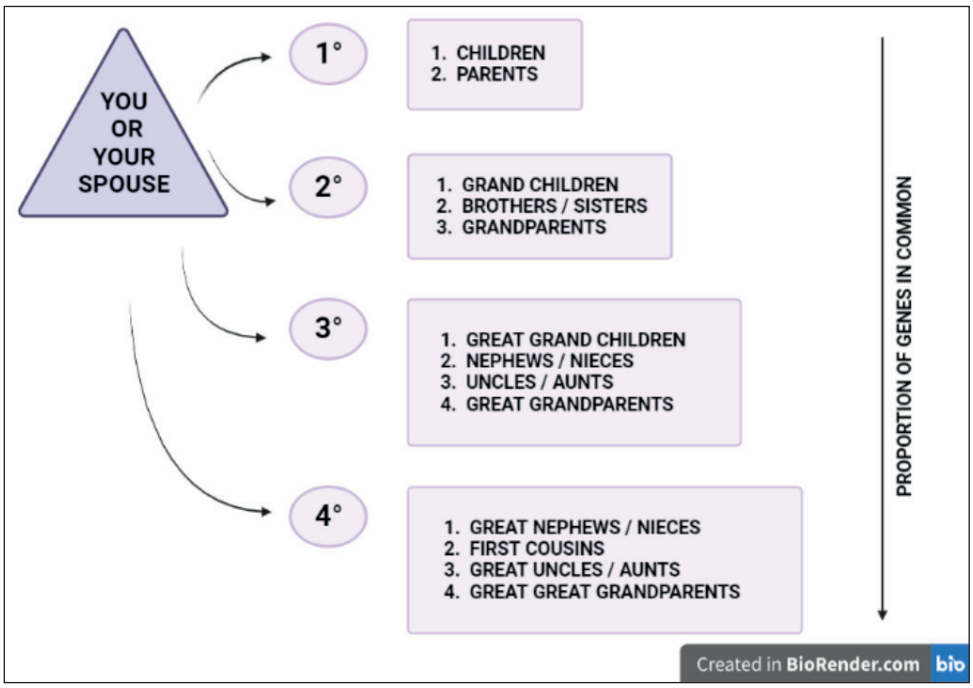

Figure 1. *Degrees of biological relationship and approximate proportion of shared genes among relatives. Figure created using BioRender.com.*

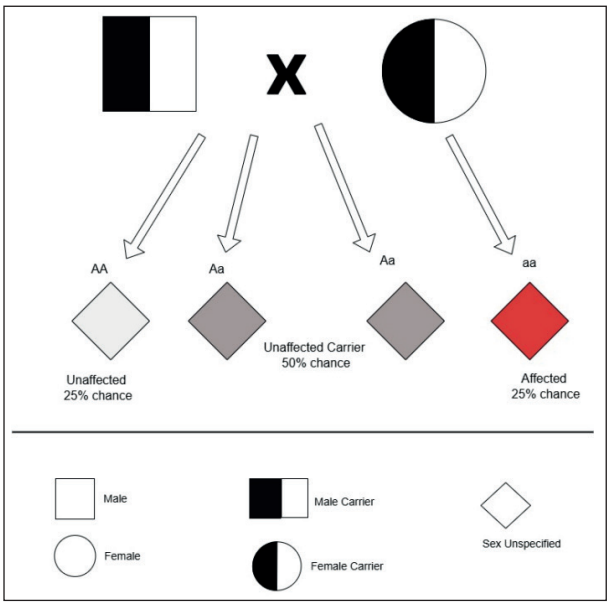

Figure 2. *Autosomal recessive pattern of inheritance from carrier parents (Aa X Aa), showing offspring probabilities - 25% unaffected (AA), 50% carriers (Aa), and 25% affected (aa). Figure created using Diagrams.net.*

equals 0.0625, reflecting increased genetic relatedness and helping to explain the higher burden of recessive disorders observed in such populations (8). Contemporary genomic studies further support this observation, demonstrating increased runs of homozygosity and a higher burden of rare deleterious variants in the offspring of consanguineous unions, directly linking consanguinity to autosomal recessive disease risk (22,23).

Epidemiological studies consistently show that the risk of congenital anomalies in offspring of first-cousin unions is modestly but significantly higher than that observed in the general population, although these risks should not be overstated or generalized across all consanguineous marriages. The baseline risk of major birth defects in the general population is approximately 2%-3%, increasing to about 4%-6% among offspring of first-cousin unions, representing an additional risk of approximately 1.7%-2.8% above the population average (8,24).

Importantly, the magnitude of genetic risk varies according to the degree of biological relatedness, the presence of shared pathogenic variants, and the genetic characteristics of the population. The increased risk associated with consanguinity is primarily attributable to autosomal recessive disorders, including certain neurodevelopmental disorders, metabolic diseases, and other monogenic conditions resulting from increased homozygosity (23–27). However, genetic outcomes vary among families because not all consanguineous couples carry the same pathogenic variants.

When both parents are carriers of the same autosomal recessive pathogenic variant, each pregnancy carries a 25% probability of producing an affected child, a 50% probability of producing a carrier child, and a 25% probability of producing an unaffected non-carrier child (28) (Figure 3). These Mendelian probabilities apply only to specific autosomal recessive conditions in carrier couples and should not be generalized to all consanguineous marriages. Consequently, assessment of genetic risk should be based on biological relatedness, family history, carrier status, and population genetic factors rather than on social or cultural classifications alone.

Consanguineous Marriages in India: Prevalence, Regional Variation, and Changing Trends

The prevalence of consanguineous marriages in India shows marked regional variation influenced by cultural, social, and demographic factors. Early anthropological studies by Beck (29) identified southern states, particularly Tamil Nadu, Andhra Pradesh, Karnataka, and present-day Telangana, as regions with higher rates of consanguinity. Subsequent studies have supported this regional pattern, with relatively lower prevalence observed in northern and north-eastern regions, although variations exist across different populations (27).

Recent data from the National Family Health Survey (NFHS-5, 2019-2021) indicate that approximately 10%-11% of marriages in India are consanguineous, with higher prevalence reported in some southern states, including Tamil Nadu, Karnataka, and Andhra Pradesh (approximately 25%-26%) (30,31).

Demographic studies indicate a gradual decline in consanguineous marriage practices over time. Sahoo et al. (32) reported a reduction in consanguineous marriages across southern states, which may be associated with broader socio-economic transitions. Kerala shows a comparatively lower prevalence of consanguineous marriages than other southern states, which may be related to differences in social development, education, and changing marriage patterns (33).

Despite this decline, considerable variation persists among different populations. Earlier studies have demonstrated that some groups historically showed higher levels of consanguinity, whereas others reported lower prevalence (33). Tamil Nadu has consistently reported relatively higher rates, although it has also experienced a substantial decline over recent decades.

This changing trend has been associated with demographic and social transitions beginning in the late twentieth century, including declining fertility rates, smaller family sizes, increased educational attainment, and greater socio-economic mobility. These factors have contributed to changes in marriage patterns, with increasing consideration of education, occupation, and broader social networks (33).

Regional and Sociocultural Patterns of Consanguineous Marriage in India

Consanguineous marriage practices in India demonstrate considerable regional and sociocultural variation. In some regions, close biological relationships in marriage have traditionally been discouraged, whereas in other regions such unions have been integrated into established kinship systems.

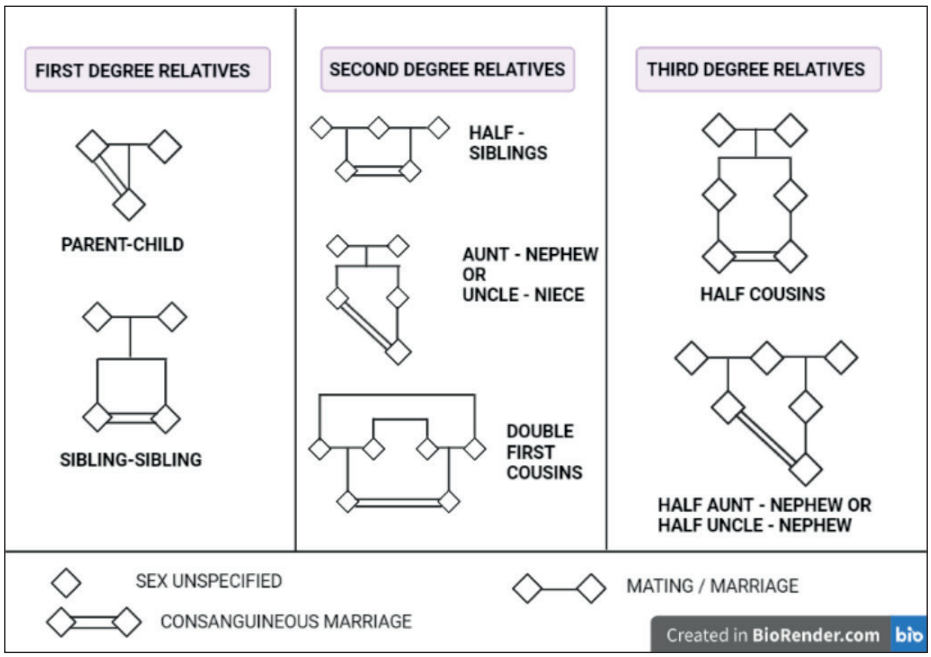

Figure 3. *Pedigree chart portraying first-, second- and third- degree consanguineous marriages and their mating patterns. Figure created using BioRender.com.*

In northern India, particularly among several Indo-European-speaking Hindu populations, marriages between close biological relatives have historically been restricted due to social and cultural norms discouraging close-kin unions (34,35).

In contrast, southern India has a long-standing history of consanguineous marriage practices, including uncle-niece and cross-cousin unions, such as marriage between a maternal uncle's daughter and a male relative. Data on cousin marriages among women aged 15-49 years indicate higher prevalence in several southern states compared with other regions, with an overall national prevalence of approximately 8% (30) (Figure 4). These practices are influenced by kinship structures and may be associated with factors such as family relationships, social compatibility, and inheritance patterns (36,37).

Consanguineous marriage is also observed in several Muslim communities in India, although prevalence varies according to region, community, and socio-cultural context (38). This variation highlights that marriage practices are shaped by diverse historical, cultural, and demographic factors rather than by a single pattern across populations.

Overall, the distribution of consanguineous marriage practices in India reflects a complex interaction of historical, social, cultural, and economic influences, demonstrating the diversity of kinship systems across the country (31,39) (Figure 5).

Genetic Counseling and Public Health Approaches to Consanguinity

The continued occurrence of inherited disorders associated with increased homozygosity highlights the importance of evidence-based genetic counseling and public health strategies. Genetic counseling provides a scientifically informed and culturally sensitive approach to communicating genetic risks, supporting informed decision-making, and respecting individual autonomy.

In contemporary India, factors such as urbanization, migration, and changing social structures have influenced marriage patterns and traditional practices. Within this changing landscape, culturally appropriate genetic counseling, premarital and preconception carrier screening, and community awareness programs can contribute to reducing the burden of inherited disorders while respecting social diversity.

Premarital genetic counseling is particularly relevant for couples with a family history of genetic disorders, known carrier status, or biological relatedness. Such counseling enables individuals to understand potential genetic risks, consider available testing options, and make informed reproductive decisions. Evidence suggests that genetic counseling improves awareness of inherited disease risk and supports informed family planning practices (40).

From a public health perspective, genetic counseling should be integrated into preventive genetic healthcare services. Carrier screening may help identify couples at increased risk of transmitting specific autosomal recessive conditions and facilitate appropriate reproductive counseling. Newborn screening programs also contribute to early identification and management of treatable inherited metabolic and genetic disorders. Establishing referral pathways involving clinical geneticists, genetic counselors, and specialized diagnostic centers is essential for individuals and families identified as being at increased genetic risk.

All genetic services should follow ethical principles, including informed consent, confidentiality, respect for autonomy, and non-directive counseling. Genetic

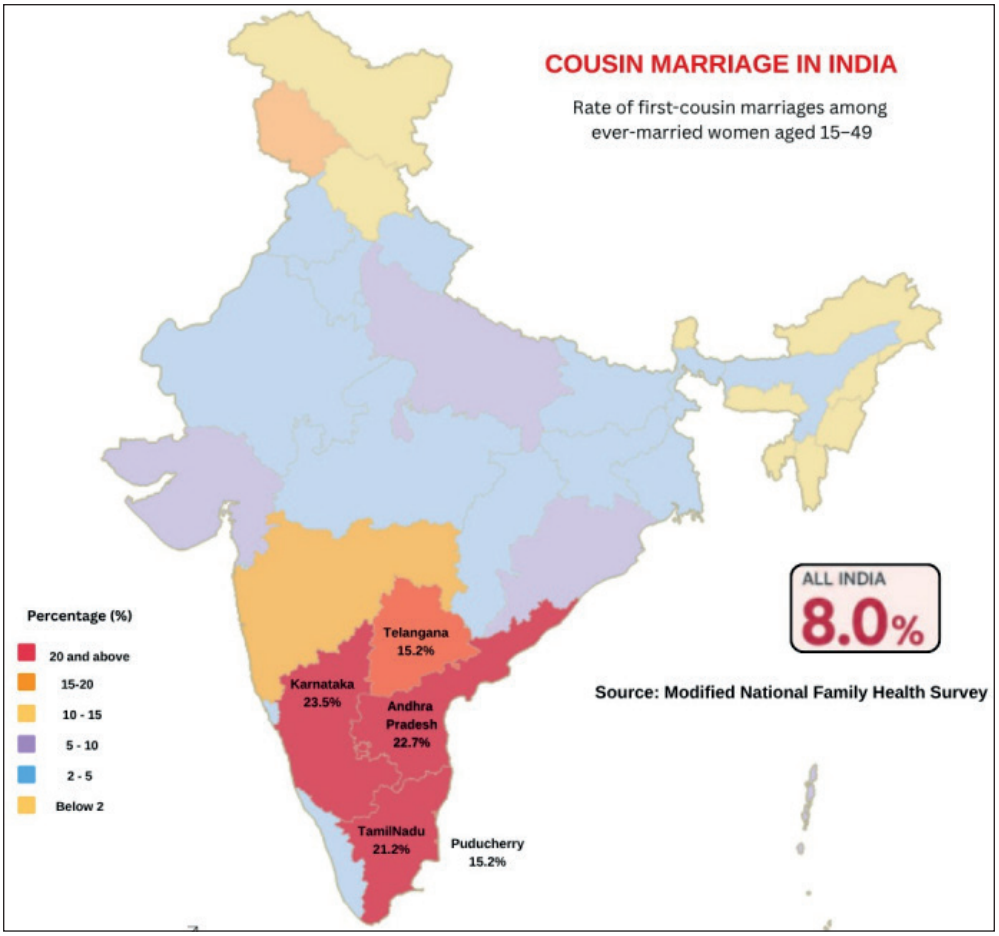

Figure 4. *Illustrates the state-wise prevalence of first-cousin marriages among ever-married women aged 15-49 years in India. The highest prevalence is observed in southern states, including Karnataka (23.5%), Andhra Pradesh (22.7%), and Tamil Nadu (21.2%), with moderate prevalence in Telangana (15.2%) and Puducherry (15.2%). The overall national prevalence of first-cousin marriage is approximately 8.0%. Data source: Modified National Family Health Survey (NFHS). Created in canva.com.*

information should be communicated clearly and sensitively, without creating stigma or influencing personal marital or reproductive choices.

Although consanguinity may increase the likelihood of autosomal recessive disorders due to shared ancestry and increased probability of carrying identical pathogenic variants, genetic risk assessment should be based on biological relatedness, family history, carrier status, and population-specific factors. Cultural lineage systems such as Gotra should not be considered genetic indicators of disease risk.

The magnitude of genetic risk associated with consanguinity should also be interpreted appropriately. Although consanguineous unions are associated with an increased probability of recessive genetic disorders, most children born to consanguineous couples are healthy. When both parents carry the same autosomal recessive pathogenic variant, each pregnancy has a 25% probability of producing an affected child, a 50% probability of producing a carrier child, and a 25% probability of producing an unaffected non-carrier child. These Mendelian probabilities apply only to specific carrier couples and should not be generalized to all consanguineous marriages. However, advice about such marriages should be given carefully, without forcing or discouraging people. The focus should be on helping individuals make informed decisions, understand possible risks, and access proper counseling support.

Genetic counseling in this context also raises important ethical considerations. Ethical considerations in genetic counseling include respecting personal and cultural values, preventing stigma, maintaining confidentiality, obtaining informed consent for genetic testing, and ensuring equitable access to genetic services. Effective counseling should provide accurate information and support informed choices while maintaining a non-judgmental and culturally respectful approach. Furthermore, ethical practice requires transparency regarding the use of genetic data for research purposes and ensuring equitable access to counseling services across different socio-economic groups (41-46).

Limitations

This review is based on previously published literature, and its findings are limited by the availability and quality of existing studies. Variations in study design, differences in cultural and demographic contexts across studies may also influence the interpretation of results.

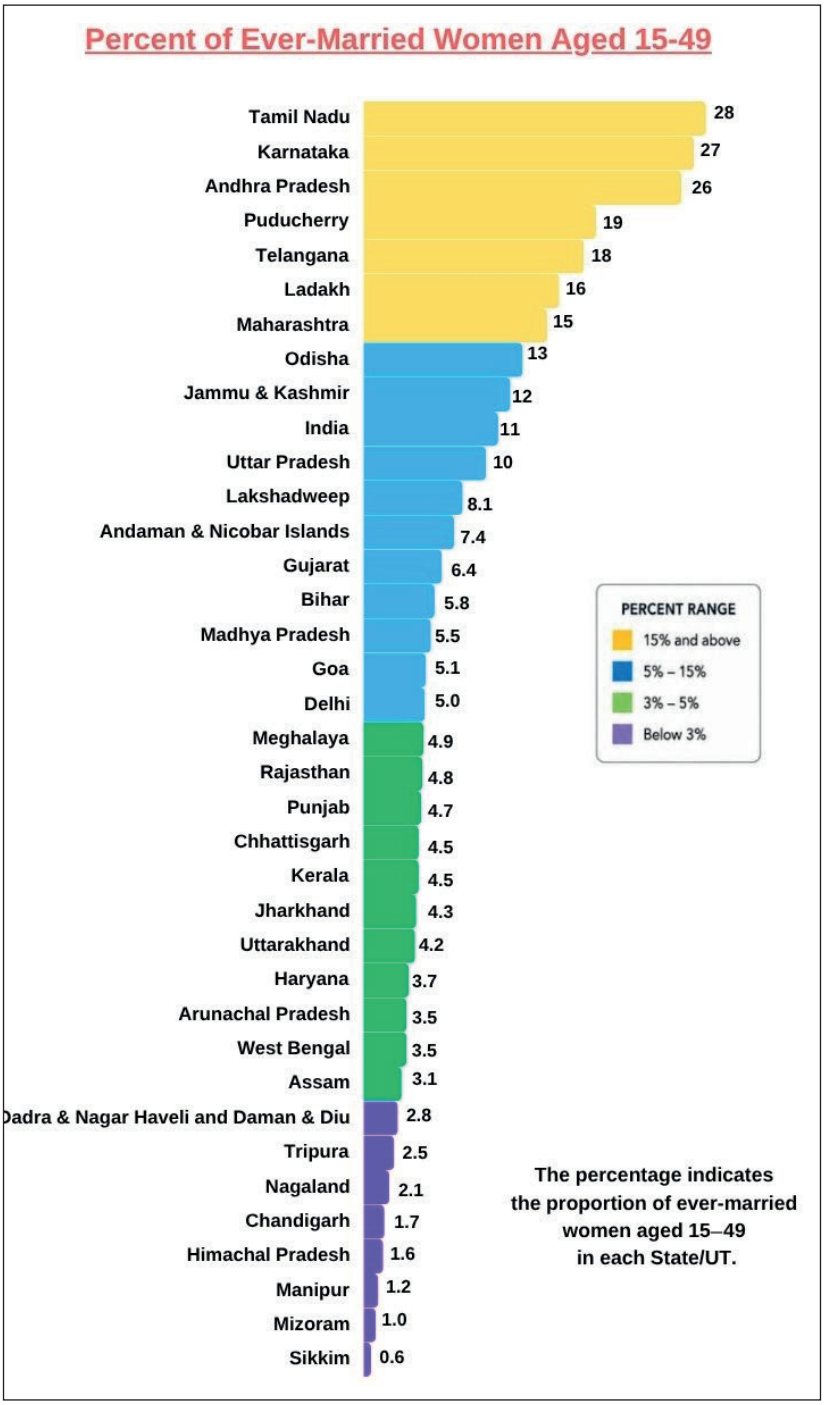

Figure 5. *Percentage of ever-married women aged 15-49 years across Indian States and Union Territories. Data source: Modified National Family Health Survey (NFHS). Figure created using Canva.*

Conclusion

The relationship between Gotra exogamy, consanguinity, and genetic disease risk highlights the need to distinguish socio-cultural traditions from biological mechanisms of inheritance. While the Gotra system has historically influenced marital practices in India, it is best understood as a cultural lineage system rather than a reliable indicator of genetic relatedness or disease risk.

Genetic risk is more accurately determined by biological relatedness, shared ancestry, family history, carrier status, and population-level factors such as consanguinity, endogamy, and founder effects. These factors can increase homozygosity and the risk of autosomal recessive disorders, and these risks are best evaluated using established principles of population genetics, genomic analysis, and genetic counseling.

From a public health perspective, culturally sensitive genetic counseling, carrier screening, premarital and preconception risk assessment, and newborn screening can help reduce the burden of inherited disorders. Integrating scientific evidence with culturally appropriate

healthcare practices provides a balanced approach to improving genetic health awareness and informed decision-making in diverse populations.

Conflicts of interest

The authors declare that they have no conflict of interest regarding the publication of this article.

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