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ANALYSING THE RISKS OF CONSANGUINEOUS MARRIAGE THROUGH PROBABILITY THEORY AND MENDELIAN GENETICS

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Abstract: This paper examines the genetic risks of consanguineous marriage using the principles of probability theory and Mendelian genetics. The study models how such unions increase homozygosity and the likelihood of inheriting deleterious recessive alleles through the application of Hardy–Weinberg equilibrium and the inbreeding coefficient F. For first-cousin marriages, the risk of recessive disorders notably rises. By integrating probability-based models and empirical data, the research quantifies genetic risks across varying degrees of consanguinity. Case studies from the Middle East, South Asia, and Africa reveal how sociocultural traditions intersect with genetic factors, underscoring the importance of culturally informed genetic counselling. The findings advocate for an interdisciplinary approach combining genetics, probability modelling, and public health to guide preventive strategies. This synthesis enhances our understanding of hereditary risk and supports evidence-based health policies in populations where consanguineous marriage remains common.

Keywords. consanguineous marriage; probability theory; Mendelian genetics; inbreeding coefficient; hereditary risk modelling.

Introduction

Consanguineous marriage (second cousins or closer) increases homozygosity through identity-by-descent (IBD), quantified by the inbreeding coefficient F [1]. For first-cousin offspring $F = 1/16$ [1]. Under inbreeding, Hardy–Weinberg proportions become

$$P(AA) = p^2 + Fpq, \quad P(Aa) = 2pq(1 - F), \quad P(aa) = q^2 + Fpq, \quad p = 1 - q,$$

which implies for a fully recessive disorder

$$P(\text{affected}) = q^2(1 - F) + qF \approx q^2 + Fq.$$

Quantitative counseling therefore hinges on correct F and allele-frequency usage; see [2, 4, 3] for clinical and public-health perspectives.

Discussion

Accurate risk communication in consanguineous unions requires correct pedigree-based F and inbreeding-adjusted genotype proportions. Single-locus absolute risks may be modest, yet

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cumulative multi-locus effects and background congenital risks justify careful, respectful counseling and, where feasible, targeted screening [4, 2].

Assumptions here include a single bi-allelic, fully recessive locus; fixed q ; no selection or de novo mutation; and no dominance leakage. Real pedigrees can contain loops and uncertain allele frequencies. Interval estimates (e.g., Bayesian models, Monte Carlo pedigrees) and genome-wide autozygosity metrics (runs of homozygosity) can refine risk [3]. Extending to multi-locus settings and structured populations is a natural direction.

Problem statement

Consanguineous marriage, defined as the union between people who share a common ancestor, has profound genetic implications that justify a rigorous analysis. This practice, although culturally prevalent in several regions of the world, particularly in parts of the Middle East, North Africa, and South Asia, exhibits a complex interplay of social, demographic and genetic factors. According to Popescu et al. (2025), in their study on the "social and demographic determinants of consanguineous marriage", the prevalence of such unions is significantly influenced by socioeconomic status, cultural traditions, and local laws, resulting in a tendency toward consanguinity that varies across populations. In specific communities, support for consanguineous marriages is based on the consolidation of family ties, maintenance of wealth, and preservation of cultural identity. However, these cultural advantages are often eclipsed by the potential genetic risks arising from such unions.

The importance of analyzing consanguineous marriages through the frames of Mendelian probability and genetics theory becomes increasingly evident when considering the hereditary risks associated with genetic disorders. As highlighted in the exploration of Mirzayeva of genetic risks and results, consanguineous unions raise the probability that descendants inherit recessive genetic features because of the greater likelihood that both parents can carry identical alleles for specific genes. This phenomenon can be modelled quantitatively using the principles of probability theory, specifically Hardy-Weinberg's balance, which is essential for understanding allele frequencies in populations.

For example, consider a scenario in which the frequency of a recessive allele (Q) in a population is 0.1. According to the Hardy-Weinberg principle, the expected frequency of homozygous recessive individuals, who would express the recessive phenotype, can be calculated using the equation q^2 . In this case, $q^2 = (0.1)^2 = 0.01$ indicating that 1% of the population would be presented with the recessive condition when considering that the unions are not sensitive. However, in a consanguineous marriage where both partners are carriers of the recessive allele, the probability that their descendants inherit the recessive feature will become $1/4$ 25%, as clarified by the Mendelian proportions. This marked increase in risk underscores the need to evaluate the genetic implications of consanguinity using empirical models based on probability theory.

In addition, the implications extend beyond individual cases, influencing public health strategies and genetic counselling practices in communities with high rates of consanguineous marriage. Awareness of high genetic risk requires a more profound commitment to Mendelian genetics, in which allele inheritance patterns can be traced through family trees or pedigree

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analysis. For example, when building a pedigree table, one can visually represent the inheritance of features and evaluate the probability that future offspring will be affected by hereditary conditions. The use of such genetic tools, combined with probability calculations, provides a comprehensive understanding of the risks associated with consanguineous marriages, thereby informing potential parents of potential outcomes. Therefore, the analysis of consanguineous marriage within the combined frames of Mendelian probability and genetics theory not only clarifies the complexities inherent to genetic inheritance but also equips interested parties with the necessary knowledge to navigate these critical family and social decisions. Consanguine marriages, defined as unions between blood-related individuals, have unique genetic challenges due to the increased probability of inheriting deleterious alleles of a common ancestor. The application of probability theory is crucial for understanding and quantifying genetic risks. More specifically, using Mendelian inheritance principles, we can calculate the probability of descendants inheriting genetic disorders that can arise from these unions.

To illustrate the genetic implications of consanguineous marriage, consider the concept of the inbreeding coefficient (F), which provides a measure of the probability that an individual inherits two alleles from any place of an ancestor common to both parents. For first-cousin unions, the inbreeding coefficient is calculated as follows:

$$F_{\text{firstcousins}} = \frac{1}{16}.$$

$$F = \sum_{A} \left(\tfrac{1}{2}\right)^{n_1(A)+n_2(A)+1}(1+F_A),$$

where the sum runs over common ancestors A (typically $F_A = 0$).

This coefficient implies that first cousins share, on average, 12.5% of their alleles identically by descent, considerably increasing the risk of recessive genetic disorders manifesting in their offspring. In addition, the risk of genetic disorders in offspring can be modelled using the Hardy-Weinberg equilibrium, which postulates that in a large population that does not experience evolutionary influences, the frequencies of genes will remain constant. In a population where consanguinity is widespread, the presence of deleterious recessive alleles can shift the balance in a non-random manner. For example, if we designate the frequency of a recessive allele q and suppose that the frequency of the associated disorder in the general population is q^2 , the probability of an offspring of consanguineous parents inheriting the disorder can be expressed as:

$$P(AA) = p^2 + Fpq, \quad P(Aa) = 2pq(1-F), \quad P(aa) = q^2 + Fpq, \quad p = 1-q.$$

$$P(\text{affected}) = q^2(1-F) + qF.$$

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For $q = 0.01$ and $F = \frac{1}{16}$,

$$P(\text{affected}) = 0.0001 \times (1 - \tfrac{1}{16}) + 0.01 \times \tfrac{1}{16} \approx 0.000719 \approx \tfrac{1}{1390}.$$

Additionally, the relationship between consanguinity and genetic disorders can be explored through case-control studies to quantify the increase in the incidence of disease. For example, in populations with high consanguinity rates, such as specific communities in the Middle East or South Asia, researchers have documented higher prevalence rates of autosomal recessive disorders than in non-consanguineous populations. Additionally, modelling the genetic risk of consanguineous marriages involves examining demographic data related to marriage patterns. Using the principles of conditional probability, we can assess how the likelihood of genetic disorders changes with various degrees of consanguinity in the population. By formulating risk assessments based on these statistical results, health professionals can better advise individuals on the genetic implications of their marital choices. The interaction between probability theory and Mendelian genetics elucidates the nuanced genetic risks associated with consanguineous marriages, providing crucial information for public health interventions and genetic counselling frameworks tailored to at-risk populations. The application of these mathematical models improves not only the understanding of genetic risks but also underlines the need for targeted educational initiatives within communities subject to consanguinity. Mendelian genetics, founded on the work of Gregor Mendel in the 19th century, outlines the principles that regulate the inheritance of traits through discrete units known as genes. Mendel's laws, in particular the law of segregation and the law of independent assortment, form the milestones of our understanding of hereditary models. In the context of consanguine marriages, in which individuals with common origins come together, the implications of these principles are particularly pronounced.

Consanguine unions increase the probability of offspring inheriting alleles identical to both parents due to shared genetic material. This phenomenon is particularly relevant for traits that show simple Mendelian inheritance, characterized by dominant and recessive alleles. For example, consider the legacy of traits such as cystic fibrosis (CF), which is governed by autosomal genes. Suppose that both parents are vectors (heterozygotes for the CF allele). In this case, they have a 25% probability of having an affected child (recessive homozygote), a 50% probability of having a carrier child, and a 25% probability of having a non-bearer child (dominant homozygote).

Inbreeding depression refers to the reduced biological form in a given population due to the accumulation of deleterious alleles, which is particularly concerning in families in which consanguinity is on the agenda. The mathematical expression for calculating the inbreeding coefficient (F) provides information on the expected increase in homozygosity. This coefficient quantifies the probability that two alleles at a locus in an individual are identical by descent from a common ancestor. For a simple model in which consanguinity between first cousins occurs, the formula can be expressed as:

$$F = \frac{1}{2} \left(\frac{1}{2} \right)^{n}$$

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Where n represents the number of generations to the common ancestor, this formula reveals that the inbreeding coefficient for first cousins is generally about 0.0625 (or 1/16), effectively translating into an increase in risk of 6.25% for the expression of recessive traits.

Given these high risks, it is essential to consider the most significant implications of consanguineous marriages on genetic health. Some populations may show a greater prevalence of genetic disorders as a direct consequence of these unions due to the presence of everyday genetic contexts, further complicating public health efforts. Population health data can often illuminate the models of variance in allele frequency, underscoring the importance of genetic counselling in communities where consanguinity is prevalent.

In summary, Mendelian genetics provides a robust picture of the mechanisms of inheritance in consanguineous marriages. By applying probabilistic models to estimate risks and drawing connections to phylogenetic histories, researchers can illuminate complex interactions between genetics, ancestry, and public health outcomes. The historical perspective offered by Teicher (2024) enriches this analysis, locating the evolution of genetic understanding in the social contexts of family structures and hereditary health risks. Consanguineous marriages, defined as unions between individuals who share a common ancestor, have unique genetic risks that can be effectively quantified and modelled through the structures of probability theory and Mendelian genetics. The transmission of alleles from ordinary ancestors increases the likelihood of homozygosity for deleterious recessive alleles, which may lead to increased health risks among children. As described by Shaw (2020), the risks associated with consanguineous unions are mainly due to the inheritance of the same genetic variants from both parents. For example, in a simple model involving biallelic loci, an individual has a probability P of transporting a harmful recessive allele given the prevalence of this allele in a population. The probability of two related individuals carrying a harmful allele can be approximated by calculating their shared genetic background using the inbreeding coefficient (F). Using the formula:

$$P(\text{affected child}) = q^2 + 2pqf$$

Where q is the frequency of the recessive allele, p is the frequency of the dominant allele, and f represents the coefficient of consanguinity, researchers can model the most likely of genetic disorders arising from consanguineous unions.

Moreover, the demographic factors influencing the health risks associated with consanguinity are multifaceted, encompassing social, cultural, and economic dimensions. Bittles (2003) highlights how Western attitudes towards consanguineous marriages often fail to understand the different implications these marriages have for public health initiatives. In many populations, the social acceptance of cousin marriages may be linked to the preservation of family lineage and cultural heritage, thereby mitigating perceived risks through community support. However, as public health campaigns seek to address associated genetic risks, it is imperative to incorporate models that not only calculate potential inheritance scenarios but also contextualize them within sociocultural structures.

To further elaborate on this modelling, consider a scenario in which two cousins marry each other. The inbreeding coefficient (F) for first cousins is 1/16, determining the likelihood of homozygosity; thus, any recessive disease is higher than in non-bleeding peers. Specifically, if

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the prevalence (Q) of a recessive disorder in the general population is 1/1000, the probability of a child being affected by this consanguineous marriage can be estimated as follows:

$$P(child\ affected) = \left(\frac{1}{1000}\right)^2 + 2\left(\frac{999}{1000}\right)\left(\frac{1}{1000}\right)\left(\frac{1}{2}\right) \approx 0.000001 + 0.000124 = 0.000125$$

Thus, the model predicts that approximately 1 in 8,000 children born to first-degree relatives can inherit the recessive disorder, significantly increasing the risk compared to non-consanguineous pairs.

These models not only provide an analytical framework for understanding genetic health risks but also emphasize the critical need for informed public health policies that consider both genetic and sociocultural variables. By leveraging the ideas of empirical research, such as Shaw (2020) and Bettle (2003), stakeholders in the domains of genetics and public health must design directed interventions that meet the exclusive needs of consanguineous marriages, thus increasing the general health results of affected populations. The implications of consanguineous marriage extend beyond the theoretical domains of probability theory and Mendelian genetics; they manifest themselves in tangible results, as observed in several case studies across different populations. Notably, Shaw and Hurst (2008) emphasized the importance of genetic counselling in informing individuals and families about the potential risks associated with such unions, particularly in culturally specific contexts in which consanguinity remains prevalent.

An illustrative case can be found in the analysis of the Alawi community in Yemen, where a high prevalence of consanguine marriages was reported. In a study analyzing genetic disorders in this population, researchers found that marriages initially significantly increased the incidence of autosomal recessive conditions. Using Mendelian genetics, the probabilities associated with the inheritance of these genetic characteristics were determined. For a child born to parents who are cousins, the risk of inheriting a genetic disorder can be determined using the following equation:

$$P = \frac{1}{4}(q^2 + 2pq)$$

Where q represents the frequency of the recessive allele in the population and p represents the frequency of the dominant allele. This calculation effectively illustrates how genetic disorders may arise from an increase in the genotypic similarity between consanguineous partners. In another case study conducted in southern Asia, where consanguineous marriages are culturally normalized, reports indicate a marked prevalence of genetic conditions such as thalassemia. By employing probability models and analyzing family segregation patterns, it has been demonstrated that affected descendant rates can be explained using the Hardy-Weinberg principle. This principle governs genetic variation in a population, allowing researchers to determine allelic frequencies and predict the risk of disease in the offspring of related individuals.

An exemplary case that demonstrates the cultural branches of genetic counselling is seen in northern Nigeria. The prevalence of consanguineous marriages in this region, particularly among the Fulani ethnic group, raises concerns about the hereditary nature of sickle cell disease.

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Genetic counselling programs have begun to educate families on the implications of intra-family marriages. Here, the following equation is emphasized, which evaluates the likelihood that children are affected by sickle cell characteristics:

$$P(s) = \frac{1}{4}$$

Where s means the haemoglobin trace in the shape of a scythe. This counselling has led to informed decisions among married couples, encouraging them to undergo premarital screening to assess the compatibility of their genotypes with the characteristics inherited from sickle cells. In addition, a study by the University of Flinders on consanguinity in Indigenous Australian populations showed an interaction between culture and genetics. The researchers found that Indigenous communities that practice consanguineous marriages exhibit high levels of genetic disorders, including cardiovascular anomalies. By using statistical models to represent genetic and selection pressures, it became evident that maintaining genetic diversity through non-consanguineous unions could mitigate the risks associated with heritable conditions.

These case studies emphasize the genetic risks associated with consanguineous marriages and the critical role of probability theory and genetic counselling in informing marriage decisions across various cultural contexts. The interaction between these factors illuminates the pressing need for structural strategies that can effectively address the genetic implications of consanguineous unions, thus promoting informed community practices and health outcomes. The analysis of consanguineous marriage through the lens of Mendelian probability and genetics theory has significant implications for understanding the genetic risks associated with such unions. Through systematic examination, it has been established that consanguinity increases the probability of homozygous recessive conditions due to the inheritance of identical alleles from common ancestors. This phenomenon can be quantified using endogamy coefficients, which provide information on the probability of offspring inheriting genetic disorders. Specifically, if individuals are first cousins, the likelihood that the offspring will be affected by a recessive genetic disorder can be approximated using the following equation:

$$P = \frac{1}{4} \cdot f$$

where p is the probability of an affected child and f is the endogamy coefficient. For first cousins, f is equal to 0.0625, which leads to an increased risk of genetic disorders. This model highlights a critical link between consanguinity and inheritance patterns, necessitating comprehensive risk assessments in populations involved in cousin marriages. Additionally, research emphasizes the significance of utilizing tools based on probability theory to predict the emergence of genetic disorders. Using Markov processes, researchers can develop sophisticated models that illustrate the generational impact of consanguineous marriages on genetic diversity. For example, considering a simplified model in which a population boycotts cousin marriage, genetic variance (denoted as var) among future generations can be expressed as

$$Var(n) = Var(0) \cdot (1 - r)^n$$

where n is the generation number and r is the relationship between the parents. This indicates that in generations in which consanguineous marriages are minimized, a systematic reduction in

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the incidence of genetic disorders can be anticipated. In light of these findings, practical implications for genetic detection are essential for future public health efforts. Currently, genetic counseling serves as a protective measure against the risks discussed. Policy formulators and health professionals must explore these implications further to develop personalized detection programs, especially in regions where consanguineous marriages are culturally normative. Recent criticisms presented by Paul and Spencer (2016) underscore the need to contextualize historical perspectives on prime marriage within contemporary frameworks, advocating nuanced discussions that balance cultural importance with genetic realities. As genetic detection practices evolve, the integration of Mendelian principles into public health strategies will become increasingly vital. Future research should focus on developing accessible programs that can efficiently deliver genetic testing services to at-risk populations. In addition, the ethical ramifications of these interventions require exhaustive examination. The statement surrounding the right to marry and the stigma associated with genetic evidence in diverse cultures requires a sensitive approach that respects family connections while prioritizing public health.

Given the ongoing developments in genetic research, it is essential to maintain interdisciplinary dialogue encompassing genetics, epidemiology, social sciences, and ethics. As awareness of the complexities of consanguineous marriage and its associated genetic risks grows, the health policy landscape can be better informed to promote not only awareness but also proactive measures to safeguard genetic health across generations. The interaction between genetics and cultural practices represents a rich area for exploration, underscoring the need for ongoing investigations in this field.

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