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Evaluation of Family-Based Ancestry Line Using Pedigree Analysis of Y-Chromosomal Inheritance Pattern – An Application to the Simulated Data

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ABSTRACT

Genetic disorders have been a health issue for a long time, and inheritance patterns are a key part of figuring out how diseases are passed down from one generation to the next. While much attention has been given to autosomal and X-linked inheritance, the role of the Y-chromosome in genetic diseases and their transmission has received far less focus. This study examines Y- chromosomal inheritance patterns, and genetic diseases to fill this gap. The simulated data contains a total of 5 different cases for different number of individuals (12, 16, 20, 24 and 33) with different penetrance. The evaluation was carried out by comparing different inheritance patterns in cases of full penetrance versus reduced penetrance. The aim of this study is to provide a fundamental practical demonstration of Y-chromosomal inheritance in families under different conditions.

Keywords: Genetic Disorders; Transmission; Pedigree; Inheritance; Autosomal; X-Linked; Y-Chromosomal; Full Penetrance; Reduced Penetrance.

1. Introduction

The study of genetic inheritance is essential for understanding the transmission of traits and diseases from one generation to the next. The foundation of this concept is known as Mendelian inheritance, which is named after Gregor Mendel [1]. It describes the predictable transmission patterns of genes, which are found on chromosomes. Chromosomes, which are formed of tightly coiled Deoxyribonucleic acid (DNA), contain the genetic information that defines an individual's traits [2]. In DNA, genes are encoded, which are the functional components of heredity that regulate specific characteristics, including physical attributes, susceptibility to disease, and the inheritance of family ancestry patterns [3].

Among the 23 pairs of chromosomes in humans, the sex chromosomes X and Y are responsible for determining the gender [4,5]. The Y-chromosome is unique that it is passed exclusively from father to son, which is the foundation of Y-chromosomal inheritance [6]. Y-chromosomes do not vary significantly from generation to generation unlike autosomal chromosomes, which undergo mutations during reproduction. This makes them a very useful tool for tracking paternal lineage and understanding family-based ancestry trends [7]. Y-chromosomal markers can show strong ancestry links, which makes this type of inheritance very useful for researching genealogy, population migration, and evolutionary history.

The Y-chromosome is important for figuring out our heritage, but it also has genes that are linked to certain male features and genetic diseases. Y-linked diseases, such as certain forms of male infertility, are passed from father to male offspring due to mutations in genes located on the Y-chromosome [8]. These disorders are a critical area of research in genetic disease studies, as they are inherited strictly in the paternal line and only affect males.

A pedigree chart is a graphical representation that displays the inheritance patterns and phenotypes of a certain gene or organism, as well as its ancestors, throughout several generations [9]. This is most typically used in the context of humans. In genetics, pedigree analysis is a key tool. The family trees can be created using the concept of pedigree to see how inheritance patterns change over generations. It advances the fields of genetics, anthropology, historical research and forensic research [10]. Y-chromosomal analysis provides useful medical insights by identifying genetic markers linked to hereditary diseases and conditions that affect male family members [11]. Pedigree analysis is an effective method for determining the male lineage within a family when combined with Y-chromosomal data. This study explores family links using pedigree charts, with a specific focus on the inheritance patterns of Y-chromosomal markers across generations.

1.1. Study Objectives

The main objectives of this study are to:

1) Provide a practical demonstration of Y-chromosomal inheritance pattern using pedigree analysis.
2) Extract the features of this pedigree analysis strategy for different conditions of full and reduced penetrance.
3) Evaluate the extracted features for different family sizes having various patterns of relations.

2. Y-Chromosomal Inheritance Pattern

The inheritance of genes on a Y-chromosome is referred to as Y-chromosomal inheritance [12]. All the male mammals carry both sex chromosomes X and Y. Since, Y-chromosomes only appear in males, Y related genes can only be passed from father to son [13]. In general, the Y-chromosome is smaller than the X-chromosome. So, it contains fewer information [14], and mostly carries the gene for determining sex. It also contains genes for sperm formation, specific hormone production, and other male-specific characteristics [15].

Usually, females typically have two X-chromosomes (XX), males have one X-chromosome and one Y-chromosome (XY) [4]. Since, the Y-chromosome is only passed down from fathers to sons, traits that are linked to genes on the Y-chromosome can only be passed down through the father's line. This means that, if a father carries a certain trait on his Y-chromosome, he will pass it down to all of his sons, and they will have the same trait. Conversely, daughters do not inherit the Y-chromosome from their fathers, and therefore do not inherit Y-linked traits [16]. Understanding Y-chromosomal inheritance helps us understand how fathers pass traits like male-pattern baldness or color blindness to sons. It traces traits through male lines in families. The main characteristics of Y-chromosomal inheritance are: only males are affected, the disease is visible in every generation of a family, a person is affected due to his father, and, an affected father is expected to have all of the sons being affected in his next generation [17].

It could be challenging to identify Y-linkage due to the fact that Y-chromosomes are smaller and contain fewer genes than X-chromosomes. The human Y-chromosome is approximately 60 mega bases (Mb) in length, and comprises only 2% of the human genome [18]. The structure of this chromosome is compact that contains approximately 62.5 million base pairs of DNA [19]. On the other hand, approximately 156 million base pairs of DNA are found in the X-chromosome [20].

3. Materials and Methods

3.1. Pedigree Data Generation

The data was generated through simulation using the R programming language. The generated data consists of three generations of a family, simulated under both conditions of full penetrance and reduced penetrance. Each individual in the dataset was assigned an identification number (ID) from 101 to 133, and a total of 5 cases for 12, 16, 20, 24, and 33 individuals were included in the data. The 6 variables of the data are: Family ID, Individual ID, Father ID, Mother ID, Sex and Disease Exposure Status. The variable " Family ID " belongs to the family number. For example, the same number is given for the individuals from the same family (Table 1) that is, the value “1” indicating that the project has been finished for a single family. The variable " Individual ID " presents a unique identification number for each individual from the same family such as 101, 102, and so on. "Father ID" and "Mother ID" are the identification numbers for the individuals (offspring, “0” in this column indicates the corresponding individual is included in this family from other family by marriage). The variable "Sex" is defined as "1" for male and "2" for female. The exposure of a disease for a particular person is presented by the variable "Disease Exposure Status" (affected=1 or unaffected=0). All the visualization and demonstration of analysis were accomplished with R programming language.

Table 1. Pedigree data for 12 individuals with full and reduced penetrance

Family ID	Individual ID	Father ID	Mother ID	Sex	Disease Exposure Status (Full Penetrance)	Disease Exposure Status (Reduced Penetrance)
1	101	0	0	1	1	1
1	102	0	0	2	0	0
1	103	101	102	1	1	1
1	104	101	102	2	0	0
1	105	101	102	1	1	1
1	106	0	0	2	0	0
1	108	103	106	1	1	0
1	109	103	106	2	0	0
1	110	103	106	1	1	1
1	107	0	0	1	1	1
1	111	107	104	1	1	1
1	112	107	104	2	0	0

4. Results

This Pedigree analysis was used to analyze all the ancestry patterns to identify how diseases are inherited from one generation to next. It allows visual inspection and identification of passing traits using pedigree data, which is crucial to ancestry analysis. Here, a total of five pedigree data cases with 12, 16, 20, 24, and 33 individuals were considered.

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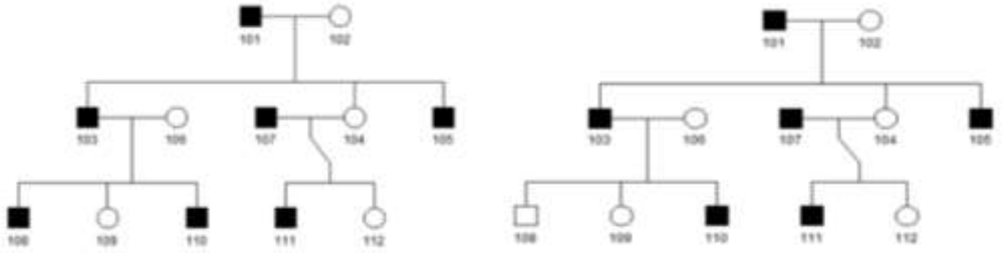
Case 1: The pedigree analysis for 12 individuals with full and reduced penetrance



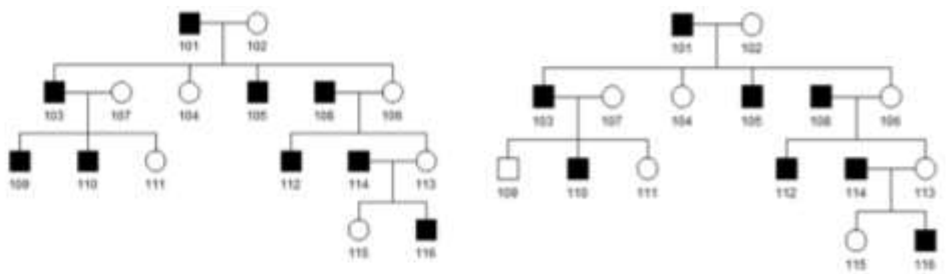
(A) (B)

Figure 1. Pedigree charts of Y-Chromosomal ancestry patterns for 12 individuals. (A) with full penetrance and (B) with reduced penetrance.

In first generation of Figure 1(A), the father (Individual ID-101) is affected but the mother (Individual ID-102) is healthy. So, all of their sons (Individual ID-103 and 105) are affected due to their father. In the second generation, an affected male (Individual ID-103) mates with a normal female (Individual ID-106) resulting their all sons (Individual ID-108 and 110) are affected. In the same generation, an affected male (Individual ID-107) mates with a normal female (Individual ID- 104) resulting their only son affected. But, none of the daughters are being affected in any generation though their fathers were affected. This indicates Y-chromosomal inheritance pattern with full penetrance. But, in the third generation of Figure 1(B) shows that the affected father (Individual ID-103) mates with a normal female (Individual ID-106), then one of their sons (Individual ID-108) being unaffected who is expected to be affected. This clearly indicates about reduced penetrance in this generation.

Full penetrance means that all people who have the genes for a trait display that trait. In Figure 1(A), all the male members have the gene for a specific trait and they expressed the trait. So, this pedigree shows and supports the full penetrance of Y-linked inheritance pattern. On the other hand, reduced penetrance occurs if some individuals who carry the gene for a trait do not express the trait. In Figure 1(B), although the affected father (Individual ID-103) passes the Y-linked gene to his sons, one son (Individual ID-108) remains unaffected. This indicates that the gene is present but not expressed, showing reduced penetrance in this pedigree.

Case 2: The pedigree analysis for 16 individuals with full and reduced penetrance



(A) (B)

Figure 2. Pedigree charts of Y-Chromosomal ancestry patterns for 16 individuals. (A) with full penetrance and (B) with reduced penetrance.

In Figure 2(A), the first generation shows an affected father (Individual ID-101) who transmits the trait to all of his sons (Individual ID-103 and 105), while their mother (Individual ID-102) is normal. This direct father-to-son

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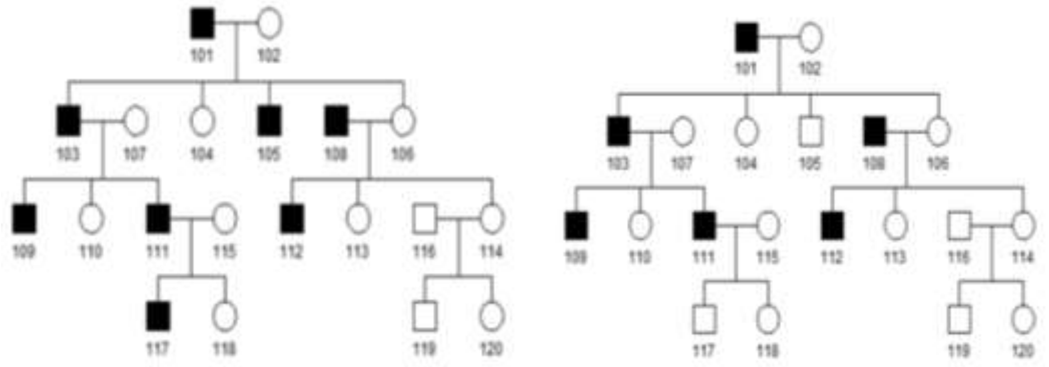
transmission is consistent with Y-linked inheritance.

In the second generation, the affected males (Individual ID-103 and 108) pass the trait to their respective sons (Individual ID-109, 110 and 112) in the third generation. The transmission pattern continues into the fourth generation, where an affected male's (Individual ID-114) only son (Individual ID-116) is also affected. Importantly, no daughters are affected in any generation. This lineage strongly indicates full penetrance of a Y-linked inheritance pattern. However, in Figure 2(B)**,** Individual ID-109 is unaffected in the third generation despite having an affected father (Individual ID-103). Under a strict Y-linked inheritance model, all sons of an affected father should inherit the trait. Therefore, this is the case of reduced penetrance**.**

Case 3: The pedigree analysis for 20 individuals with full and reduced penetrance



(A) (B)

Figure 3. Pedigree charts of Y-Chromosomal ancestry patterns for 20 individuals. (A) with full penetrance and (B) with reduced penetrance.

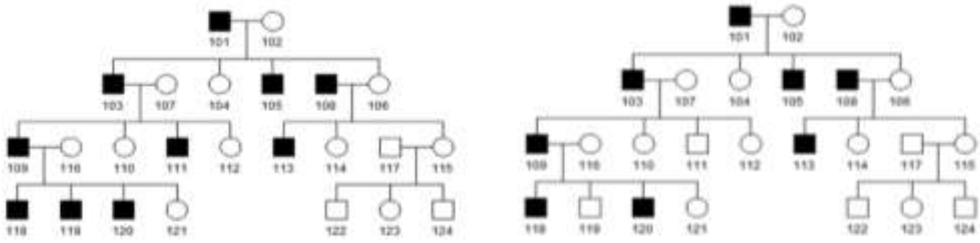
According to Figure 3(A), the father (Individual ID-101) exhibits symptoms of the disease, while the mother (Individual ID-102) displays no such symptoms. Consequently, all the male offspring (Individual ID-103 and 105) inherit the disease in the second generation because of their affected father.

Moving to the second generation, an affected male (Individual ID-103) mates with an unaffected female (Individual ID-107), resulting all the male offspring (Individual ID-109 and 111) inheriting the disease in the third generation. Similarly, an unaffected female (Individual ID-106) marries with an affected male (Individual ID-108) from another family, leading to their only male offspring (Individual ID-112) being affected in same generation.

Again, in the third generation, a female (Individual ID-115) mates with an affected male (Individual ID-111), resulting their only male offspring (Individual ID-117) inheriting the disease. Furthermore, a mating between two unaffected parents (Individual ID-114 and 116) results all the healthy offspring, irrespective of gender. The observation that all male members possess the gene for a specific trait and exhibit the corresponding trait indicates the full penetrance of the Y-linked inheritance pattern.

But, Figure 3(B) shows that Individual ID-105 (second generation) and Individual ID-117 (fourth generation) are unaffected while their fathers (Individual ID-101 and 111; in the first and third generation) are affected. It indicates reduced penetrance in disease passing history.

Case 4: The pedigree analysis for 24 individuals with full and reduced penetrance



(A) (B)

Figure 4. Pedigree charts of Y-Chromosomal ancestry patterns for 24 individuals. (A) with full penetrance and (B) with reduced penetrance.

In the first generation of Figure 4(A), an affected father (Individual ID-101) transmits the trait to all of his sons (Individual ID-103 and 105). This is a clear father-to-son transmission, while females remain unaffected, is the characteristic of Y-linked inheritance.

In the following generations, this pattern continues in a similar fashion. The affected male (Individual ID-103), when mates with an unaffected female (Individual ID-107), passes the trait to all of his sons (Individual ID-109 and 111). Similarly, in another case of same generation, an affected male (Individual ID-108) transmits the trait to his only son (Individual ID-113), despite mating with an unaffected female (Individual ID-106). In both cases, transmission occurs strictly from father to son, and no daughters are affected.

In the third generation, the affected male (Individual ID-109) transmits the trait to all of his sons (Individual ID-118, 119 and 120). In contrast, when both parents are unaffected (Individual ID-115 and 117) none of their children male or female are being affected. The consistent father-to-son transmission and complete absence of affected females across all generations strongly support a Y-linked inheritance pattern with full penetrance as shown in Figure 4(A).

However, Figure 4(B) shows a deviation from this strict pattern. In the second generation, although an affected male (Individual ID-103) mates with a normal female (Individual ID-107), one of their sons (Individual ID-111) remains unaffected. The same is true for Individual ID-119 in the fourth generation. Under a fully penetrant Y-linked model, all sons of an affected father should inherit the trait. Furthermore, in the fourth generation, the sons (Individual ID-122 and 124) of the unaffected father (Individual ID-117) are also unaffected. This interruption in the expected father-to-son transmission indicates reduced penetrance in the family history.

Case 5: The pedigree analysis for 33 individuals with full and reduced penetrance

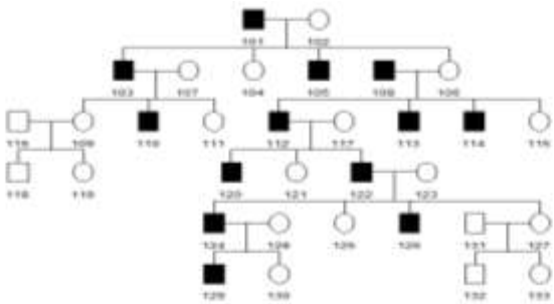


Figure 5. Pedigree chart of Y-Chromosomal ancestry pattern with full penetrance for 33 individuals.

In the first generation, an affected father (Individual ID-101) transmits the traits to all of his sons (Individual ID-103 and 105) in the second generation. The exclusive father-to-son transmission, with no mention of affected females, is consistent with a Y-linked inheritance pattern (Figure 5).

In the third generation, the pattern continues without interruption. The affected male (Individual ID-103), when mated with an unaffected female (Individual ID-107), passes the traits to his only son (Individual ID-110). Similarly, an unaffected female (Individual ID-106) mates with an affected male (Individual ID-108), and all of their sons (Individual ID-112, 113, and 114) inherit the traits. In both family branches, in the third generation, transmission occurs strictly from father to son, and no daughters are affected.

In the same generation, an affected male (Individual ID-112) transmits the traits to all of his sons (Individual ID-120 and 122) after mating with an unaffected female (Individual ID-117). In contrast, two unaffected parents (Individual ID-116 and 109) have none of their children are being affected regardless of gender as the father is healthy.

In the fourth generation, the affected male (Individual ID-122) passes the traits to all of his sons (Individual ID-124 and 126) in the fourth generation. Finally, in the fifth generation, the affected male (Individual ID-124) transmits the traits to his only son (Individual ID-129), maintaining the uninterrupted father-to-son inheritance.

Across all generations, every affected father transmits the trait to all of his sons, no daughters are affected, and unaffected parents do not have affected offspring. This consistent paternal transmission without exception strongly confirms a Y-linked inheritance pattern with full penetrance in this pedigree.

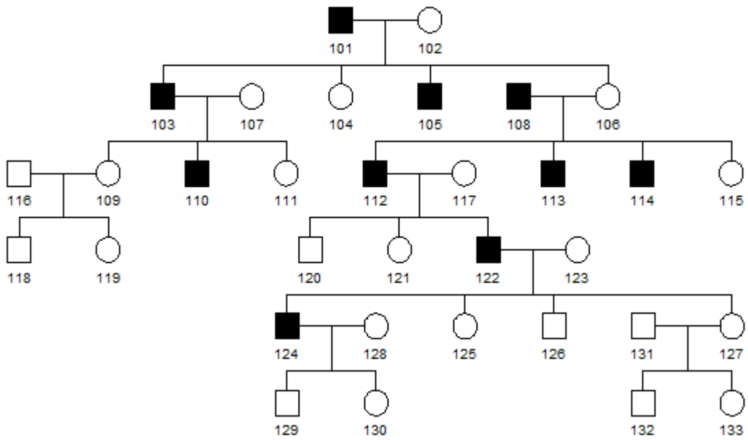


Figure 6. Pedigree chart of Y-Chromosomal ancestry pattern with reduced penetrance for 33 individuals.

In Figure 6, the inheritance pattern shows deviation from a strict Y-linked transmission and can be interpreted generation by generation. Individual ID- 120 is expected to be an affected person due to his affected father (Individual ID-112). But he is healthy due to reduced penetrance.

In the fourth generation, an affected male (Individual ID-122) mates with a normal female (Individual ID-123). Under a typical Y-linked inheritance model, all sons of an affected father should inherit the trait. However, one of their two sons (Individual ID-124) is affected while another (Individual ID-126) remains unaffected. This incomplete father-to-son transmission contradicts the expectation of full penetrance.

In the next generation (fifth), the affected male (Individual ID-124) mates with a normal female (Individual ID-128). According to strict Y-linked inheritance, their only son (Individual ID-129) should also be affected. However, he remains unaffected, further interrupting the expected paternal transmission pattern.

Although the trait appears only in males and follows a paternal line, the presence of unaffected sons born to affected fathers clearly indicates reduced penetrance in this pedigree.

5. Discussion

This study focuses on the development and description of pedigree charts concerning Y-chromosomal inheritance patterns. The simulated data was generated using the R programming language under both conditions of full and reduced penetrance. The visualization of these ancestry patterns is displayed via the respective pedigree for both the full penetrance and reduced penetrance patterns.

It provides a detailed explanation of how diseases are passed down from parents to offspring and focuses on the processes of data generation and computation. It discusses how Y-chromosomal inheritance differs from other modes of genetic inheritance with a focus on the unique transmission pattern of traits carried by the Y-chromosome.

This paper includes the structure of pedigree data and in-depth explanations of family charts for both full penetrance and reduced penetrance. There is also a comparison of the pedigree charts for full penetrance and reduced penetrance.

Y-chromosome inheritance patterns are very important in genetic studies because they help us understand how traits and genetic disorders are passed down. Y-chromosome markers are used to find paternal lineages and to study population genetics and the history of evolution. Genetic professionals have a vital role in determining the genetic risk linked to Y-chromosomal inheritance patterns. They can predict the possibility of inheriting Y-linked illnesses and counsel affected people and families by analyzing family pedigrees and genetic data.

6. Conclusion

This study demonstrates the use of pedigree charts to analyze Y-chromosomal inheritance patterns using simulated data generated in R. By illustrating both full penetrance and reduced penetrance, the report highlights how Y-linked traits are transmitted exclusively through the paternal line and how penetrance variability influences phenotype expression and complicates pedigree interpretation. The findings emphasize the importance of Y-chromosomal markers in lineage tracing, genetic risk assessment, and population studies. Finally, this study focuses on a practical demonstration of Y-chromosomal ancestry analysis. Despite of simulated data, it could be a practical guideline for the introductory researchers.

7. Future Directions

This study could be further extended to:

1) Apply the analysis process for the high dimensional real data.

2) Calculate the individual risk of disease exposure using traditional probabilistic approach.

3) Predict the risk by the Bayesian approach.

8. Limitations

This study uses the simulated data due to the insufficient funding. Also, the lab does not have enough tools to manage the large amounts of sequencing data that could be obtained from a few data storage websites.

Declarations

Source of Funding

This study did not receive any grant from funding agencies in the public, commercial, or not–for–profit sectors.

Competing Interests Statement

The authors declare no competing financial, professional, or personal interests.

Consent for publication

The authors declare that they consented to the publication of this study.

Authors' contributions

Both the authors took part in literature review, analysis and manuscript writing equally.

Informed Consent

Not applicable for this study.

Availability of data and material

All supplementary documentation can be provided for the purpose of academic inquiry or verification.

Institutional Review Board Statement

Not applicable for this study.

Ethical Approval

Not applicable for this study.

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