



INHERITANCE PATTERN AND ASSOCIATION STUDIES OF SOME HUMAN MENDELIAN TRAITS AMONG DIFFERENT COMMUNITIES FROM NAGPUR, INDIA.

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ABSTRACT:

Physical qualities are passed down from parents to offspring as traits. Freckles, blood type, hair colour, and skin tone are all examples of characteristics. Mendelian attributes are traits that are passed down through one gene's dominant and recessive alleles. Alleles are alternate form of genes, which are basically bits of DNA that carry information about a particular trait. Non-Mendelian traits are those that are not dictated by dominant or recessive alleles and they can be caused by more than one gene. From December 2016 to March 2017, this research was carried out. The methodology employed in this study was a blend of descriptive and interviewing techniques, as well as personal observations. In this study we have looked at the morphogenetic traits of Earlobe Attachment, Tongue Rolling, widows peak, Morton's Toe, Hitchhiker's Thumb, Eye color, Hair colour in relation to genotype in different communities.

Key words: - *Inheritance pattern, Mendelian traits, communities.*

INTRODUCTION:

All living creatures are a product of long evolutionary life process on earth. Every organism shares this history. Humans are curious about their ancestors and wonder about the process that led to their origin. It's really difficult to understand the details about human origin, of which genetic study explains us a lot.

A specific segment of DNA, called gene, determines observable characteristics also known as physical traits. Nucleus of cell encloses all the genes for the organisms survival and are arranged on particular chromosomes. All homologous chromosomes (except X and Y) contain two copies (a pair of alleles) of each gene. The X and Y also have a few loci carrying homologous or allelomorphic genes. Each parent contribute one copy of each gene at the time of conception. These original genes (allele) store the

information needed for the cell to assemble proteins which actually yields specific physical traits.

Genes exhibit two or more variables, called alleles. The offspring may inherit two identical or two different alleles from their parents. Two different alleles when present interact in specific ways. An inherited trait, for example, a physical feature, a blood group or an enzyme system, may depend on a single pair or on a cumulative effect of a large number of genes. The former is called Mendelian or unit factor inheritance and the later polygenic inheritance (Kennedy, 2005). Qualitative (monogenic) traits are the classical Mendelian traits such as form (eg: round and wrinkled seeds), structure (horns or hornless condition in cattles), pigments (black and white coat colour) and antigens and antibodies (blood group) and so on.

Each qualitative characteristic may be genetically controlled by two or more alleles of a single gene, with little or no environmental changes to conceal the gene effects.

The organisms possessing qualitative traits have distinct phenotypic classes and are said to exhibit discontinuous variation.

Genes carried on chromosomes are responsible for the development of inherited characters or traits. When a single gene pair determines a trait, the trait is known as a single gene trait and its mode of inheritance from one generation to the next follows Mendel's law of unit factor inheritance and segregation.

The Mendelian or unit factor inheritance may be either autosomal or sex linked. In autosomal trait, the genes are located on any of the 22pairs of autosomes and in sex linked traits, the genes belong to the X and Y chromosome.

An inheritance is called autosomal dominant when one member of an allelic pair is able to express itself in total disregard for the presence of the other member. On the other hand, autosomal recessive inheritance depends on the expression of both partners of an allelic pair. When the two members of an allelic pair are identical, they are said to be homozygous and when they are unlike each other, the combination is said to be heterozygous. A dominant gene expresses itself in both homozygous and heterozygous forms, whereas a recessive gene only expresses itself in homozygous form.

A trait is said to constitute the phenotype of an individual, while the allelic pair of genes determining the trait constitutes the genotype of that trait. The traits presented here are determined by single gene. However it may be possible that some may involve more than one gene. (Speicher, M. R., & Antonarakis, 2010).

MATERIALS AND METHODS:

The present survey has been carried out to investigate morpho-genetical status of Muslim, Christian, Buddhist, Hindu and Sikh communities of Nagpur. The survey is preliminary attempts to determine the possible combination patterns that exist between expressed morphogenetic traits. Our focus in this case is on following parameters

- I. Blood group system
- II. Earlobe Attachment
- III. Tongue Rolling
- IV. Widow's peak
- V. Morton's Toe
- VI. Hitchhiker's Thumb
- VII. Eye colour
- VIII. Hair colour

In this survey simple random sampling technique was adopted in choosing subjects. Consent was sort and given by subjects.

RESULT & DISCUSSION:

I. Blood group system.

Blood groups are antigens found on the membranes of red blood cells that are coded by alleles at different loci on the chromosome. Despite this, about 400 blood grouping antigens have been identified. The most significant are ABO and Rh. The ABO blood group genes are located on chromosome 9q34.2. Diverse geographical and ethnic groupings have different ABO and Rh(D) blood groups. ABO type has been the single most significant test conducted in blood banking services to avoid mortality since its discovery by Landsteiner in 1900.

Individuals are grouped into four major blood types based on the antigens found on RBC: A, B, AB, and O. The human RBCs that contain antigen D are known as rhesus positive (Rh*), while those without antigen D in their RBCs are rhesus negative (Rh*) (Daniels *et al.*, 1995, Schenkel-Brunner, 2000,).

Several investigations on the distribution of blood types among various endogamous populations in India have been conducted during the last few decades (Bhasin et al. 1992, 1994, Bhasin and Walter 2001). For the present study blood samples were taken from finger pricks, and open slide method of testing for ABO blood groups and Rh (D) factor was followed (Bhasin and Chahal 1996). ABO Grouping and Rhesus The allele frequencies for these two systems were calculated according to the method of Mourant et al. (1976).

II. Earlobe Attachment:

If earlobes hang free they are detached. If they attach directly to the side of the head, they are attached earlobes. Few scientists have reported that this trait is governed by single gene. The Unattached earlobe is a dominant trait and attached earlobe a recessive trait. The most common form of ear lobe is hang free, which is a dominant (EE or Ee) feature, and the less common attached ear lobe is recessive (ee) (Williams and Hughes 2005, Ahmed and Yaas, 2013).

III. Tongue Rolling:

In 1940, the famous geneticist Alfred Sturtevant noted that about 70 % people of European ancestry were able to roll up the lateral edges of the tongue, while the remaining 30% were unable to do so. Several investigators have studied this attribute (Abimbola, 2019, Odokuma et al., 2008 Koomson, 2020)

The ability of rolling the tongue may be the expression of a single gene. The ability to roll the tongue is considered a dominant trait and the inability to do so a recessive trait (Hsu, 1948). However inheritance of this trait remain to be in question as scientists observed that around 30% of identical twins do not share the trait.

IV. Widow's peak:

The rare 'V' shape hairline in the centre of forehead in some people is called 'Widow's peak'. It is genetically a dominant trait. One parent if show

presence of widow's peak and the other do not, the possibility is 50% of their children's inherit widow's peak. If one parent possesses both alleles dominant then the percentage will be 100. Kyriakou et al., 2021 pointed out that though Widow's peak is a usually overlooked, yet it is a significant morphogenetic trait.

V. Morton's Toe:

It is the condition of a shortened first metatarsal in relation to the second metatarsal. It is a type of brachymetatarsia. The metatarsal bones behind the toes vary in length. For most feet, a smooth curve traced through the bases of the toes but in Morton's foot, the line has to bend more sharply to go through the base of big toe. Exercises or the use of a flexible pad beneath the first toe and metatarsal may be used as conservative treatments for Morton's toe pain.

VI. Hitchhiker's Thumb:

The ability to hyperextend the thumb (extend it backwards at the last joint) is due to a autosomal recessive allele. A straight thumb is dominant to Hitchhiker's Thumb.

VII. Eye colour:

In humans, pigmentation of the eye varies from light brown to black, depending on the concentration of melanin in the iris pigment epithelium (located on the back of the iris), the melanin content within the iris stroma (located at the front of the iris), and the cellular density of the stroma. Humans and other animals have many phenotypic variations in eye colour.

The genetics of eye colour is complicated, and colour is determined by multiple genes. So far as many as 15 genes have been associated with eye colour inheritance.

VIII. Hair colour:

Hair colour is determined by two forms of melanin: eumelanin and pheomelanin, which pigments hair follicles. Generally when more eumelanin is present, the colour of hair is darker; if less eumelanin is present the hair colour is lighter.

4. Data collection

In this study, five communities are considered. And, random sample of size 60 is taken from each of five different communities'. The frequencies of dominant and recessive genes corresponding to different traits is counted and are shown in the following tables-1.

Chi square Test for Homogeneity of Proportions (Odetunmbi *et al.*, 2020, Turhan, 2020):

This test is applied to test if different populations have the same proportion of individuals with some characteristic. This test is applied for different traits one by one starting with the trait Blood group

Application of Chi square Test for Homogeneity of Proportions for the trait blood group:

Let P_1 denote the proportion of people with respect to the Mendelian trait of Blood group (dominant) in Muslim community

Let P_2 denote the proportion of people with respect to the trait dominant Blood group in Christian community

Let P_3 denote the proportion of people with respect to the trait dominant Blood group in Buddhist community

Let P_4 denote the proportion of people with respect to the trait dominant Blood group in Hindu community

Let P_5 denote the proportion of people with respect to the trait dominant Blood group in Sikh community

$H_0 : P_1=P_2=P_3=P_4=P_5$ vs $H_1 : \text{The population's proportions are not all equal}$

Chi square = $4/42 + 4/42 + 4/42 + 4/42 + 4/42 = 0.476$

Table value of chi square with 4 d f at 5 % l s = 9.488

H_0 may be accepted at 5 % l s

Conclusion – The population proportion of people w r to the trait Blood group is same across all the five communities.

Similarly carrying out the analysis for all the mendelian traits like tongue roll, Hitchhker's

thumb, Morton's toe, Widow's peak and ear lobe, it is found that

- 1) W r to the traits Blood group, tongue roll and Hitchhker's thumb, Null hypothesis is accepted. That means the proportion of people with the mendelian traits dominant Blood group, tongue roll and Hitchhker's thumb is same across the different communities .ie it is independent of community whereas
- 2) W r to the traits Morton's toe, Widow's peak and ear lobe, Null hypothesis is rejected. That means the proportion of people with the Mendelian traits dominant Morton's toe, Widow's peak and ear lobe is not same across the different communities i.e. it is dependent on community.

5 Chi square test of independence of attributes:

This test is used to test whether the two attributes Mendelian traits and community are independent. In order to apply this test, first of all the observed frequencies shown in table 1,2 3 4 and 5 are grouped together in one table as shown below in table 8.

The Null hypothesis and the alternative hypothesis are set as follows

H_0 : Mendelian trait and community are independent vs.

H_1 : Mendelian trait and community are not independent

The study of genetics has revealed that distinctions exist in humans, which may be due to natural selection, migration, or method of inheritance, and that no two people are precisely alike (Bhasin *et.al.* 1992,).

When the genetic basis of a Mendelian phenotype is discovered, a causal relationship between genotype and phenotype is established, allowing for carrier and population screening. Morphogenetic traits can be used to look into variances that develop within and between populations as a result of genetic differences.

According to the results of the current study, no two people are same.

They differ in morphological characteristics. Genetic and environmental factors have a role in these variances. Traits differed from person to person, according to the survey. Between 40 and 90 percent of the population has blood group system, earlobe attachment tongue rolling, widow's peak, morton's toe, hitchhiker's thumb, eye colour, hair colour. These traits are known as high frequency traits because they occur frequently in the population. The rate of frequency is a statistical phrase that describes how frequently an inherited trait appears.

Many fields, such as forensic pathology, anthropology, and other related fields, rely heavily on the ability to link morphogenetic features.

CONCLUSION:

Statistical analysis shows that:

- i. The population proportion of people w r to the trait Blood group is same across all the five communities.
- ii. The proportion of people with the mendelian traits dominant Blood group, tongue roll and Hitchhker's thumb is same across the different communities .i.e. it is independent of community whereas,
- iii. W r to the traits Morton's toe, Widow's peak and ear lobe ,Null hypothesis is rejected .That means the proportion of people with the mendelian traits dominant Morton's toe, Widow's peak and ear lobe is not same across the different communities i.e. it is dependent on community .
- iv. It may be concluded that there is relationship between community and color of eye .
- v. It may be concluded that there is relationship between community and colour of hair.

From the above data & analysis, it is concluded that the two attributes, Mendelian trait and community are dependent. This means that there is association between the two attributes.

ETHICAL APPROVAL AND CONSENT

The study protocol was reviewed and approved by the Departmental Board ethical clearance committee. Before data collection, an informed consent was obtained and collected data were kept confidential.

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COMPETING INTERESTS:

Author has declared that no competing interests exist.

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Table 1 - Muslim community

Different Traits in Muslim community	Dominant	Recessive	Total
Blood group	40	20	60
Tongue Roll	44	16	60
Widow's peak	26	34	60
Ear lobe	20	40	60
Hitchhker's thumb	50	10	60
Morton's toe	32	28	60

Table 2 - Christian community

Different Traits in Christian community	Dominant	Recessive	Total
Blood group	40	20	60
Tongue Roll	50	10	60
Widow's peak	52	08	60
Ear lobe	48	12	60
Hitchhker's thumb	56	04	60
Morton's toe	54	06	60

Table 3 - Buddhist community

Different Traits in Buddhist community	Dominant	Recessive	Total
Blood group	44	16	60
Tongue Roll	30	30	60
Widow's peak	08	52	60
Ear lobe	48	12	60
Hitchhker's thumb	56	04	60
Morton's toe	56	04	60

Table 4 - Hindu community

Different Traits in Hindu community	Dominant	Recessive	Total
Blood group	44	16	60
Tongue Roll	49	22	60
Widow's peak	16	44	60
Ear lobe	12	48	60
Hitchhker's thumb	50	10	60
Morton's toe	08	52	60

Table 5 - Sikh community

Different Traits in Sikh community	Dominant	Recessive	Total
Blood group	40	20	60
Tongue Roll	56	04	60
Widow's peak	58	02	60
Ear lobe	56	04	60
Hitchhker's thumb	58	02	60
Morton's toe	14	46	60

Table 6 - Community and Eye color

Name of community	Black Eye colour	Brown Eye colour	Total
Muslim	56	04	60
Christian	42	18	60
Buddhist	28	32	60
Hindu	52	08	60
Sikh	50	10	60
Total			300

Table 7- Community and Hair color

Name of community	Black hair colour	Brown hair colour	Total
Muslim	56	04	60
Christian	58	04	60
Buddhist	40	20	60
Hindu	48	22	60
Sikh	12	48	60
Total			300