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SHORT COMMUNICATION

## Hypothesis of Selected Mendelian Traits Among Random Asian Population

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### ABSTRACT

This discusses the prospects for understanding the genetic basis of few selected traits in humans. Phylogenetic analysis provides a path to identify genes and biological processes underlying any trait influenced by inheritance. We suggest that different experimental approaches should be undertaken for traits caused by common genetic variants versus those arising from rare genetic variants. We discuss the intellectual foundations of genetic of Mendelian traits in humans by examining the degree of freedom of four particular factors , namely toung rolling (1.307), hitchhiker's tumb (3.22), widow's peak (44.825) and mid digital hair (92.825).

**Keywords:** Toung rolling, Hitchhiker's thumb, Widow's peak, Mid digital hair

### 1. INTRODUCTION

Gregor Mendel is called the father of modern genetics because his experiments with pea plants gave us the basic insights and vocabulary to accurately study genetic patterns. He used statistical analysis of large populations of plant offspring to identify all possible expressions

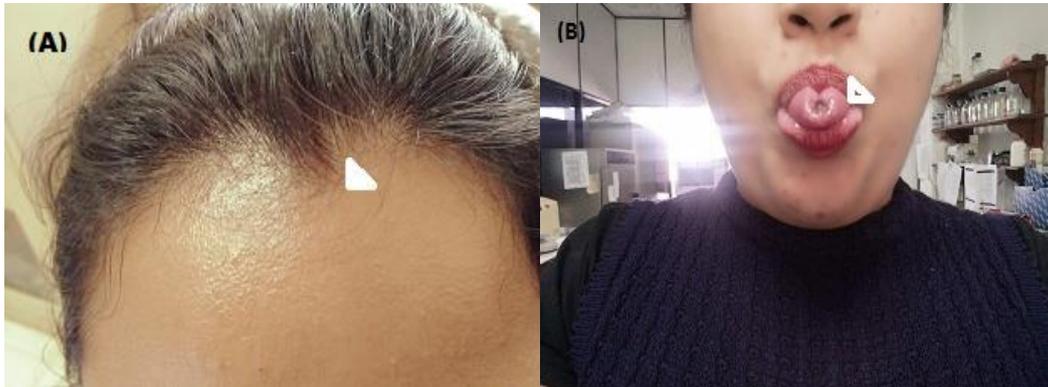
of basic genetic traits. The traits that Mendel tracked through generations of pea plants were simple dominant or recessive patterns. According to geneticists, a Mendelian trait is due to a single gene that follows classic Mendelian transmission. Instead, at conception we all inherited several distinct strands of DNA that are packaged within a complex system of proteins. Hence, it is the chromosome and not the gene that is truly the physical unit of inheritance. A gene is usually defined as a section of DNA that contains the blueprint for a polypeptide chain. The term locus carries with it the implication that a gene has a fixed location on a chromosome. A single human chromosome will contain several thousands of genes.

The phenotypic traits of the different organisms may be of two facts, namely qualitative and quantitative. The quantitative traits are also called metric traits. They do not show clear cut differences between individuals and forms a spectrum of phenotypes which blend imperceptively from one type to another to cause continuous variations. In contrast to qualitative traits, the quantitative traits may be modified variously by the environmental conditions and are usually governed by many genes, each contributing such a small amount of phenotype that their individual effects cannot be detected by Mendelian methods but by only statistical methods. Such genes which are non-allelic and effect the phenotype of a single quantitative trait, are called polygenes. The inheritance of poly genes or quantitative traits is called quantitative inheritance, multiple factor inheritance, multiple gene inheritance or polygenic inheritance.

The genetical studies of qualitative traits are called qualitative genetics. Certain Characteristics of Quantitative Inheritance The quantitative inheritance have following characteristics: The segregation phenomenon occurs at an indefinitely large number of gene loci, If a substitution of a allele occurs in a gene locus then such allelic substitutions have trivial effects. The genes for a multiple trait have different biochemical functions but similar phenotypic effects, therefore, the phenotypic effects of gene substitutions are interchangeable. Blocks of genes are bound together by inversions and transmitted as units from inversion heterozygotes to their progeny, but such blocks are broken up by crossing over in inversion homozygotes. The polygenes have pleiotropic effects; that is, one gene may modify or suppress more than one phenotypic trait. A single allele may do only one thing chemically but may ultimately affect many characters. The environmental conditions have considerable effect the phenotypic expression of poly genes for the quantitative traits.

## **2. INHERITANCE PATTERNS IN HUMANS**

A large number of human traits which are ranging from seemingly unimportant traits such as whether one's hair forms a widow's peak (Figure1) to genetic disorders that can cause debilitating disease and death. Many of these traits are readily observable and have served as teaching tools and textbook examples for decades. For example, eye color, the ability to bend back the thumb nearly 90 degrees (known as hitchhiker's thumb), the ability to roll ones tongue into a U-shape (Figure 1), or whether one's the formation of hair at the mid digit of the finger and hitchhiker's thumb (Figure 2) were all thought to have simple mendelian inheritance patterns. Even though many people still portray these traits as having simple Mendelian inheritance characteristics such as dominant and recessive alleles of a single gene, the inheritance patterns of most of these traits are in fact much more complex.



**Figure 1.** (A) widow's peak (B) the ability to roll ones tongue into a U-shape



**Figure 2.** (A) Hitchhiker's thumb (B) Non Hitchhiker's thumb (c) Mid digital hair in two different selected ones.

Even some traits thought to have somewhat more complicated but still predictable inheritance, such as male pattern baldness (originally thought to be a sex-linked trait passed from the mother's father to her sons), turn out to be determined by much more complex interactions of multiple genes and the environment.

### **3. HARDY-WEINBERG EQUILIBRIUM (HARDY-WEINBERG PRINCIPLE)**

Hardy-Weinberg principle is used to compare allele frequencies in a given population over a period of time. A population of alleles must meet five rules in order to be considered in equilibrium: 1) No gene mutations may occur and therefore allele changes do not occur. 2) There must be no migration of individuals either into or out of the population. 3) Random mating must occur, meaning individuals mate by chance. 4) No genetic drift, a chance change in allele frequency, may occur. 5) No natural selection, a change in allele frequency due to environment, may occur. Hardy-Weinberg Equilibrium never occurs in nature because there is always at least one rule being violated. Hardy-Weinberg Equilibrium is an ideal state that provides a baseline against which scientists measure gene evolution in a given population. The Hardy-Weinberg equations can be used for any population; the population does not need to be in equilibrium.

### **4. POPULATION GENOTYPES AND ALLELES**

The Hardy-Weinberg principle applies to individual genes with two alleles, a dominant allele and a recessive allele. A population with such a gene can be described in terms of its genotype numbers, the number of individuals with each of the three resulting genotypes - or in terms of the three genotype frequencies. The frequency of each genotype is the number of individuals in the population with that genotype divided by the total number of individuals in the population, as displayed in Table 1.

**Table 1.** Genotype numbers and genotype frequencies in a hypothetical population. (Note that total genotype frequencies sum to 1.0.)

Genotypes	Number with these Genotypes	Genotype Frequencies
AA	18	0.36
Aa	24	0.48
aa	8	0.16
Total	50	1

Calculating Allele Frequencies can also describe a population, somewhat more abstractly, in terms of its allele frequencies. The frequency of an allele is defined as the total number of copies of that allele in the population divided by the total number of copies of all alleles of the gene. We can calculate population allele frequencies from genotype numbers.

The total number of dominant A alleles in our population equals 50, which is the sum of: - The number of AA individuals times 2 (the number of A alleles per individual) =  $18 \times 2 = 36$  - the number of Aa individuals (times 1, the number of A alleles per individual) +  $24 = 50$

**5. RANDOM MATING AND THE HARDY-WEINBERG PRINCIPLE**

Godfrey Hardy’s and Wilhelm Weinberg’s insight was that when a population is in equilibrium, genotype frequencies can be calculated from allele frequencies. The key insight is that with random mating, the probability that each parent transmits a given allele to an offspring is equal to that allele’s frequency in the population. Suppose the frequency of a dominant allele A in the population is p (0.60 in our example above) the frequency of a recessive allele a in the population is q (0.40 in our example above) Table 2 shows that probability of each of the possible offspring genotypes when the population is in equilibrium. (Note for purposes of calculation clarity, the table divides the heterozygous Aa offspring into two subsets, those who inherit the dominant allele from their mother and those who inherit the dominant allele from their father.

**6. METHODS FOR ANALYZING THE INHERITANCE PATTERNS**

The human genome contains ~21,000 genes that encode proteins. Each gene sits in a specific location on a chromosome. Humans have 22 pairs of autosomes and one pair of sex chromosomes (XX for females and XY for males). Clinical cytogeneticists are scientists who study chromosomes and detect and analyze hereditary diseases and abnormalities with the help of pedigrees and karyotypes. Molecular tests can also determine the presence of alleles linked to diseases and genetic variations that increase a person's chances of developing a disease. Pedigrees can be used to follow a trait through the generations. Although it is easy enough for scientists to study the genetics of model organisms such as mice and pea plants, researchers cannot study human genetics by performing controlled crosses on human subjects. Instead, researchers examine patterns of inheritance in an existing population through pedigree analysis. During Queen Victoria’s lifetime, it was known that diseases like hemophilia .

**7. DATA COLLECTION**

Inheritance Patterns	No. of individuals with dominant traits (D+H)	No. of individuals with recessive traits (R)	Total No.of individuals
Tounge rolling	34	16	50
Mid digital hair	17	33	50
Widow’s peak	8	42	50
Hichhiker’s tumb	7	43	50

The selected inheritance Patterns in humans are tongue rolling, Hichhiker’s thumb ,Mid digital hair and widow’s peak among the randomly selected Indian and Sri Lankan population. Each four characters have been observed in the each individuals.

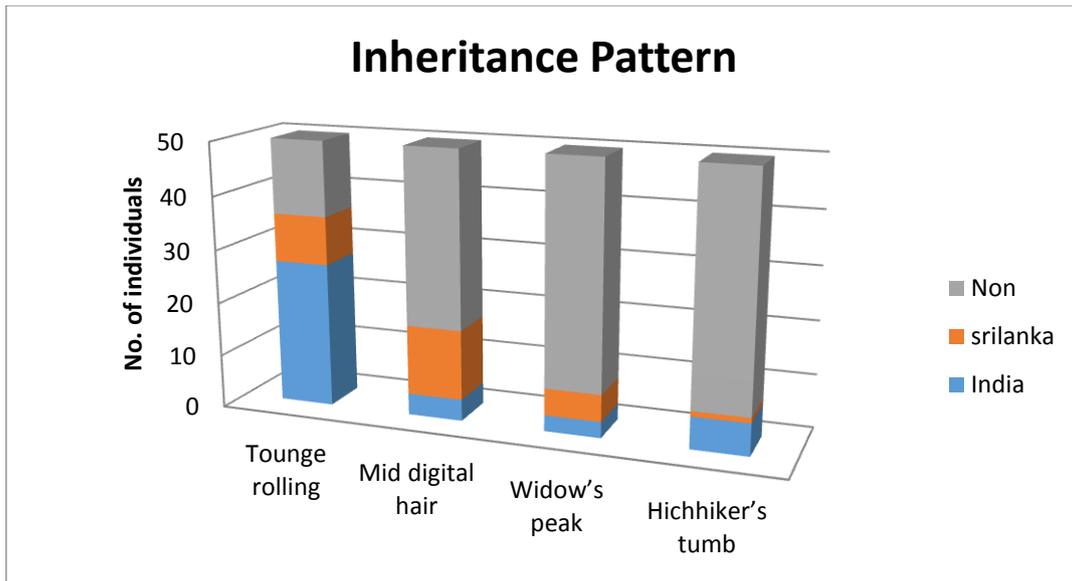
### 8. STATISTICAL DATA ANALYSIS AND RESULTS

Hypothesis of each inheritance patterns were calculated by using statistical method.

Inheritance Patterns	Genotype	Observed (o)	Expected (E)	d(O-E)	d <sup>2</sup>	d <sup>2</sup> /E	Hypothesis
Tounge rolling	Tounge rolling	34	37.5	-3.5	12.25	0.375	1.307
	Non -Tounge rolling	16	12.5	3.5	12.25	0.98	
Mid digital hair	Mid digital hair	43	37.5	5.5	30.25	0.8	3.22
	Non -Mid digital hair	7	12.5	-5.5	30.25	2.42	
Widow’s peak	Widow’s peak	17	37.5	20.5	-420.25	33.62	44.826
	Non -Widow’s peak	33	12.5	20.5	420.25	11.2	
Hichhiker’s tumb	Hichhiker’s tumb	8	37.5	-295	870.25	23206	92.826
	Non -Hichhiker’s tumb	42	12.5	29.5	870.25	69.62	

Simultaneously the dominant phenotype of tongue rolling ,hichiker’s tumb ,mid digital hair and widow’s peak were 0.327, 0.8, 33.62 and 23.206 while the recessive was 0.98, 2.42, 11.2 and 69.6. The degree of freedom is 1 for tongue rolling and hichhiker’s tumb while 65 mid digital hair and 90 for widow’s peak.The hypothesis for tongue rolling, hitchhiker’s thumb, middigital hair and widow’s peak was 1.307, 3.22, 44.825 and 92.825.

Graphical analysis of inheritance patterns



**9. CONCLUSIONS**

According to the observed and selective certain population, chi square value is 1.03 for tounge rolling. In this case the degree of freedom is 4.00 according to the chi square table, whilst it can be catergorised as null hypothesis. The other traits are mid digital hair and its chi square value is 44.825 with degree of freedom 65 and widow's peak is 92.826 with the degree of freedom 90 according to the chi square table, which is known to be alternative hypothesis does not following the hardy weinburg equilbriam. Calculated chi square value for hitchhiker's thumb is 3.22 with the degree of freedom 1, following hardy weinburg equilbriam (null hypothesis). The final observation through the Sri Lankan and Indian population, tounge rolling and hichihikers tumb is popular character among the selective population while mid digital hair and widow's peak is rare. The tally marks are the fifth and sixth genotypic preparation common among the Sri Lankan population. The fourteenth character is popular among the Indians who came from upper part of India.

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