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Understanding the Basic Principles of Population Genetics and its Application

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Abstract
This paper examined the basic issues and applications of population genetics for clear understanding of the topic. Population genetics is a new field in science and as a result, many students find the concept difficult. Major areas treated included: definition of population genetics, gene pool, gene frequency, genetic equilibrium, genetic drift, natural selection, isolation and Hardy and Weinberg principle among others. Attempts were made to treat these listed concepts in relation to evolution because population genetics provided a great support to the idea of organic evolution. Ideal population as proposed by Hardy and Weinberg was treated and disruptive influences like mutation, natural selection etc that can affect it to cause evolution was also treated. Application of Hardy-Weinberg principle was discussed which helped to determine whether a population is in equilibrium or not and at the same time helps to predict the nature of a population in the next generation. Aspects of mating systems in a population that is, random mating and non random mating (like exogamy, endogamy etc) were also examined which are important socio-culturally, especially in terms of marriage. The students of biological sciences will benefit from the contents of this paper for understanding the topic.
Keywords: Definition of population genetics, concepts in population genetics, ideal population, Hardy-Weinberg principle, mating systems in a population, evolution.

1.Introduction

Population is defined as total number of organisms of the same species living together in the same habitat. We have populations of each of human beings, dogs, orange trees, etc. Genetics is the study of heredity and variation. Heredity is the transmission of characters from parents to offsprings through the genes. Genes are discrete units found on the chromosomes that determine heredity. Chromosomes are thread-like structures, made of protein and found in the nucleus of a cell on which the genes are located. Genes usually occur in pairs on homologous chromosomes; in this case they are called alleles. Allele is one of two or more alternative forms of a gene e.g. tall / short (Tt), Red / white (Rr) etc. The ABO blood group in man has three alleles (A, B and O); height has two alleles 'T' for tall and't' for short in a heterozygous situation etc (Ramalingam, 2000; Ugwuadu, 2011). Variation is defined as differences found in organisms of the same species. For example, some organisms of the same species like man can be tall, short, black, white etc while they have different blood groups, different Rhesus factors etc. Variations of the first category like height are continuous variation because they can be affected by the environment while the second category e.g. blood group is discontinuous variation because they cannot be affected by the environment among other reasons. These explanations are fundamental in understanding population genetics (Ugwuadu, 2011). Population genetics can literally be defined as the study of heredity and variation of a given population. In its specific terms, population genetics is the study of gene taken as a unit of evolution (Fullick,1994). This definition explains that the genetic makeup of a population evolves over-time. Verma and Agarwal (2012) defined population genetics as the genetical studies for the inheritance of phenotypic traits in a given population. Population genetics has provided great support to the idea of organic evolution because various aspects of evolution can be reviewed in terms of population genetics (Verma & Agarwal, 2012). Evolution is a permanent change in gene or allelic frequencies in a population. When the change occurs, there is emergence of new species. The change can be morphological / structural, anatomical or physiological etc. Evolution is therefore, a generation-to-generation change in a population's frequencies of alleles (Nuffied, 1971; Ugwuadu, 2011).

2.1 Some Key Concepts in Population Genetics

The following concepts are explained in this paper; they include: gene pool, gene frequency, genetic equilibrium, genetic drift, natural selection, isolation, Hardy-Weinberg law, ideal population and mating systems in a population.

2.1.1 Gene Pool: Gene pool is the sum total of all the genes in a population at a given time. The gene pool of a particular trait or character is studied and not for all traits at the same time. For example, the gene pool for height is studied in all its variations among organisms of the same species. The genetic frequencies of the parental generation form the gene pool from their gametes. The gene pool is the

reservoir from which the next generation draws its genes (Campbell, Reece & Simon, 2004)

2.1.2 **Gene Frequency:** Gene frequency is the number of times any given gene occurs in a population relative to all its alleles at the same locus on the homologous chromosomes. Gene frequency is a statement of how often a gene occurs in a gene pool of a population.

2.1.3 **Genetic Equilibrium:** When gene frequencies remain constant or unchanged, there is stability called genetic equilibrium. When there is genetic equilibrium the total number of alleles remains unchanged. If two alleles should occur in equal proportions in a large isolated breeding populations and neither had a selective or mutational advantage over the other, they would be expected to remain in equal proportion and in equilibrium generation after generation. Only factors like natural selection, mutation, chance, migration etc can alter the equilibrium of two or more alleles (Roberts, 1976).

2.1.4 **Gene Flow:** Gene flow is the movement or transfer of genes as individuals migrate from one place to another and interbreed. Gene flow occurs when members of a population inter breed, genes recombine in various ways and spread in a population. Recombination of gene takes place during meiosis. Gene flow causes evolution because the nature of the gene determines the nature of the offspring. If the gene is mutated, incidentally the offspring inherits the character expressed by it which results in the emergence of new species (evolution). A population may gain or loose alleles by gene flow which is genetic exchange with another population (Campbell, Reece & Simon, 2004).

2.1.5 **Genetic Drift:** Genetic drift is a significant drop in gene frequency especially in small populations. A drop in gene frequency may cause evolution. This is when individuals carrying that particular gene reproduce unsuccessfully resulting in total loss of that particular gene. Usually a significant proportion of gene is lost to cause a drop in gene frequency resulting in the emergence of new species (evolution). The bottleneck effect and founder's effect are two situations that cause genetic drift due to chance. Bottleneck effect is caused by disasters such as earthquakes, floods, droughts and fires which may reduce the size of the population. The small surviving population may not be the representatives of the original population's gene pool. The founder's effect is a genetic drift in a new colony when a few individuals colonize an isolated island, lake or some other new habitat. The smaller the colony, the less its genetic makeup will represent the gene pool of the larger population from which the colonists emigrated (King,1995; Campbell, Reece & Simon, 2004)

2.1.6 **Natural Selection:** Natural selection is the theory of survival of the fittest propounded by Charles Darwin. The theory states that individuals that are best adapted in the environment will survive while those that are not well adapted will die or become eliminated. The reason is that all living organisms are constantly involved in a struggle for existence. In a population, those organisms that tend to survive and reproduce are those individuals whose variations give them competitive advantage over the rest. They are the fittest because they are the best adapted in the environment. The favourable variations they possess are passed on to their offspring but organisms that cannot compete are eliminated due to low adaptive features. Environmental pressure is the main cause of natural selection which may result in evolution. Favourable environmental pressures like food, water, absence of disease, weather etc may cause favourable variations to accumulate over time and unfavourable ones tend to diminish if initial pressures are unfavourable (Roberts, 1976). Over many years in many generations, favourable variations like hybrid vigor naturally selected by the environment accumulate sufficiently to give rise to new species from ancestral species (survival of the fittest). According to Charles Darwin, the theory of natural selection / survival of the fittest explained that natural selection acting through environmental pressures cause evolution. The implication of this theory to population genetics is that if conditions in the environment are favourable individuals will survive and also mate to produce healthy offspring from generation to generation. But if conditions in the environment are unfavourable, some individuals will gradual diminish or die because they lack competitive advantage like hybrid vigour. For example, the biggest, fastest, toughest frog in the pond has a Darwinian fitness of zero if it is sterile. Production of fertile offspring is the major score that counts in natural selection (Starr, 2003).

2.1.7 **Isolation:** Isolation is an important factor contributing to the process of evolution. It is the separation of population of a particular species into smaller units which prevent interbreeding between them. As a result of this separation, organisms develop new characteristics. For example, if members of a species are separated into two or more populations by natural barriers like oceans, seas, rivers, glaciers, mountains etc for a long time, the situation may result in the two separated units becoming incompatible because the barriers prevented interaction between them. Gene flow is affected because the two groups may not interbreed due to the fact that a reproductive barrier has been created (geographical isolation). Genes may change due to environmental pressures among the two groups. Apart from geographical isolation, there is isolation by time caused by evolutionary history that a population was a descendant of an original ancestors. Isolation by distance (spatial isolation) which produces changes in structure,

colour, size, habits due to species living long distances away from each other. The species may differ from one region to the other e.g. Wrens (birds) of South America (Verma and Agarwal, 2012).

2.1.8 Mutation: Mutation is a spontaneous or sudden change in the genetic make-up of an individual resulting in the formation of new characteristic (evolution). As a result of this sudden change, a new stable genetic information occurs that gives rise to distinct characteristics in a population. When the new characters are passed on to offsprings through the gametes, evolution has occurred. Mutation on body cells or somatic cells is not inheritable, but mutation that causes chromosomal aberration is inheritable because it results in change in genes (point mutation) or change in chromosomal number (Starr, 2003).

2.1.9 Hardy-Weinberg Law (HWL)

The stable gene frequencies were proved mathematically by G.H. Hardy, a British mathematician and W. Weinberg, a German physician in 1908. The law states that provided there are no disruptive influences such as mutation, natural selection etc, the frequency of genes in a population remains constant from generation to generation (Fullick, 1994). The algebraic equation developed by Hardy and Weinberg describing a stable gene frequency is:

$p^2 + 2pq + q^2 = 1$ where;

p2 = frequency of homozygous dominant individual in a population.

2pq = frequency of heterozygous individuals

q^2 = frequency of homozygous recessive individual

1 = a constant = 100% of a population (Fullick, 1994)

2.1.10 Explanation of HWL

The frequency of a particular allele in a population is a statement of how often the allele occurs and this is expressed as a decimal fraction of 1 (where 1 represents 100% of the population). There is a general formula which can be used to represent the frequency with which the dominant and recessive forms of an allele occur in the gene pool of a population. The general formula is p + q = 1 (Fullick, 1994) where letter p represents the frequency of dominant allele and q represents the frequency of recessive allele. This means that the frequency of dominant allele plus frequency of recessive allele equals to one (1) or 100%. This simple equation is of very limited value as it stands, because it can only be used to measure the frequencies of genes and not observable phenotypes. In order to solve this deficiency, Hardy and Weinberg independently developed an equation for stable genotypic frequencies within a population which can be solved using observable phenotypes and the equation is $p^2 + 2pq + q^2 = 1$ which had earlier been stated and explained. The Hardy-Weinberg formula can be used to estimate the frequency of a harmful allele which is useful information for public health programme dealing with genetic diseases (King, 1995).

2.1.11 How Hardy and Weinberg arrived at the Formula

Suppose the alleles A,a are present in a particular population for a particular trait, their frequencies are p and q for dominant and recessive characters respectively. Therefore p + q = 1 (using the general formula for calculating gene frequency).

Individuals in a population can be one of the following;

- Homozygous for the dominant allele = AA genotype
- Double recessive for the allele = aa genotype
- Heterozygous for the allele = Aa genotype

Assuming that mating is random and that all the individuals produce approximately equal numbers of gametes and the alleles A, a do not mutate, the gametes will fuse as follows using punnett square or checker board to illustrate the crosses as in figure 1.

Gametes	A	a
A	AA	Aa
a	Aa	aa

2.2 Figure 1: Crossing the Parental Genotypes Aa and Aa

The progeny / offspring arising from the crosses are AA, Aa, Aa, aa. Substituting their frequencies with p and q since p stands for homozygous dominants and q for homozygous recessive, we have;

AA = $p \times p = p^2$

Aa = $p \times q = pq$

Aa = $p \times q = pq$

aa = $q \times q = q^2$

Adding the products obtained, this gives; $p^2 + pq + pq + q^2 = p^2 + 2pq + q^2$

If the population obtained is stable or at equilibrium this gives; $p^2 + 2pq + q^2 = 1$ or $(p + q)^2 = 1$. Where 1 = 100%

of a population, hence, the Hardy and Weinberg equation for a stable frequency. Hint: Information on Gregor Mendel's principles of inheritance showing monohybrid crosses first and second filial generations are the source of explaining the Hardy – Weinberg Principle as in fig. 1.

2.3 Application of Hardy – Weinberg Equation / Equilibrium / Formula

Question 1: Tongue rolling is caused by dominant allele. Suppose in a population 84% of the people can roll their tongues where as the remaining 16% cannot. Using the Hardy – Weinberg formula, determine how many people are heterozygotes and how many people are homozygous for tongue-rolling (Robert, 1978).

2.3.1 Solution:

Let R represent the gene for tongue rollers

Let r represent the gene for non tongue rollers

Tongue rollers can be homozygous dominant (RR) or heterozygous rollers (Rr). Non-tongue rollers are homozygous recessive (rr). The genotypes of both groups will be distributed according to Hardy-Weinberg equation / formula thus;

$p^2 + 2pq + q^2 = 1$

p^2 = frequency of homozygous dominant individuals (RR)

2pq = frequency heterozygous individuals (Rr)

q^2 = frequency of homozygous recessive (rr)

But 16% of the population (q^2) are non-rollers = rr (given)

$q^2 = 0.16 \quad \left[{}^{16}/_{100} \right]$

$q = \sqrt{0.16}$

q = 0.4

but p + q = 1 (general formula for calculating frequency of genes / alleles).

p + 0.4 = 1 (substitution)

p = 1 – 0.4

= 0.6

2pq = 2 x 0.6 x 0.4 = 0.48 (substitution)

(heterozygote)

% of heterozygous individual = 0.48 or 48%

Since the % of tongue rollers in the population = 84% (given) and heterozygous individual rollers = 48%

Homozygous dominant rollers = 84% - 48% = 36%.

The genotypic frequency of the population is summarized as follows; RR Rr rr

36% 48% 16% = 100% or 1

The genotype of the next generation will also be 100% or 1 since there was no disruptive influences like mutation, selection etc in the parental generation hence the Hardy-Weinberg law is obeyed. One major problem of finding gene frequency is that, it is not possible to distinguish between the homozygous dominant and the heterozygous individuals based on their physical appearances. This means that both of them have similar phenotypes. However, using the Hardy-Weinberg equation the gene frequencies of each can be calculated from the number of homozygous recessive individuals in the population.

Hardy-Weinberg equation was developed to describe a situation of stable equilibrium where the relative frequencies of the alleles and genotypes stay at equilibrium over time. The implication is that in the absence of any factor which affects the equilibrium e.g. mutation, natural selection etc the gene frequencies remain the same in the population from generation to generation. This statement is only true in an ideal population which Hardy-Weinberg put as follows (Fullick, 1994; Verma & Agarwal, 2012).

- There is random mating; there are no factors at work to cause the choice of mates to be non random.
- The population size is large
- The population is isolated so that there is no exchange of genetic material with other populations so no migration (emigration or immigration).
- There are no mutations.
- No natural selection takes place, in other words all alleles have the same level of reproductive advantage or disadvantage. Evolution tempers with ideal population, so it may or may not exist because the gene pool equilibrium is upset.

2.4 Mating Systems in a Population (MSP)

This is a pattern of choosing mates in a population. In a population, mating or sexuality can be random or non-random / selective mating.

2.4.1 Random Mating (RM)

In a random mating, any one individual of one sex is equally likely to mate with any individual of the opposite sex. This also means that the likelihood that any two individuals in a population will mate is independent of their genetic makeup (Fullick, 1994). For example, the pollen from a wind – pollinated grass flower is likely to blow on to any one of thousands of other similar grass flowers. In a random mating, considering Rh factor (Rhesus factor) an individual is either Rh^- or Rh^+ phenotypes, random mating is crossed as follows in respect to Rhesus factor.

$Rh^+ \times Rh^-$
$Rh^- \times Rh^-$
$Rh^+ \times Rh^+$

If mating is random, the above three matings are possible. The meanings of the above mating are; Rhesus positive mating with Rhesus negative ($Rh^+ \times Rh^-$); Rhesus negative and Rhesus negative ($Rh^- \times Rh^-$) ; and Rhesus positive mating with Rhesus positive ($Rh^+ \times Rh^+$).

2.4.2 Non-Random Mating (NRM)

Non-random mating is a type of mating which is not random and several mating types fall within this category. The mating is usually selective. Non-random mating occurs when some features of the phenotype affect the probability of two organisms mating. There are some traits where mating is not random in Nigeria e.g. albino, the physically handicapped. Also animals in which the male displays in some way to attract the female do not show random mating e..g male peacock and female peacock. In human beings, non-random mating is the normal practice because individuals select their partners considering cultural and social set ups (Fullick, 1994).

Some examples of non-random mating are;

- Inbreeding (endogamy)
- Exogamy (outbreeding)
- Isophenogamy
- Heterophenogamy (Ramalingam, 2000; Ugwuadu, 2011; Verma & Agarwal, 2012). Each of these non-random mating types will briefly be explained.

2.4.3 Inbreeding (endogamy)

This is the mating of close relatives over and above that which is expected in random mating. Matings which are classified as inbreeding are; selfing, parent-child, sib-sib, uncle-niece, uncle-aunt and cousins.

Selfing is the simplest form of inbreeding. With respect to one pair of alleles A, a, there are three types of mating that come under this category of selfing;

(a) AA x AA
(b) Aa x Aa
(c) aa x aa

When homozygous dominants are selfed or crossed with one another, all the offsprings or progeny produced will be homozygous dominants like in (a) above. In the same way when homozygous recessives are crossed / selfed as in (c) above, all the offspring will be homozygous recessives. When heterozygotes are selfed or crossed with one another as in (b) above, the genotypes of the offsprings will be ¼ AA, ½ Aa, ¼ aa. Selfing reduces the frequency of heterozygotes and at the same time increases the frequencies of the two homozygotes (a) and (c) above. Parent – child mating, sib mating and cousin mating may produce the same effect as selfing, but uncle – niece mating and uncle – aunt mating may decrease heterozygote proportion to some extent because they are more distant than other inbred matings. In inbreeding, if dominance involves (c) above only the recessive homozygote will increase and alternative dominant phenotype will decrease. Many unfavourable traits are inherited as recessives e.g. sickle-cell anaemia, colour blindness, haemophilia etc hence society and law are against close endogamous marriages or what is referred to as consanquinous marriage (close blood marriage). If for example, two close relations are heterozygous for sickle cell anaemia (As), this implies that some offsprings from these parents will inherit sickle cell anaemia and this continues from generation to generation due to inbreeding. The major advantage of inbreeding is to maintain and build-up certain desirable traits in a gene population. However, some of the disadvantages of inbreeding are; in many species of plants and animals where there is close inbreeding, the offsprings become weak and may die because of hereditary diseases; there is also general lowering of resistance to diseases and decrease in fertility (Ramalingam, 2000; Ugwuadu, 2011; Verma & Agarwal, 2012).

2.4.4. Exogamy (Outbreeding)

Exogamy refers to the system of mating in which mating occurs between unrelated or distantly related individuals. This mating occurs more often than expected under the system of random mating. The genetic consequence of exogamy is an increase in heterozygosis. If dominance is involved outbreeding will result in an increase in the proportion of dominant phenotypes and a corresponding decrease in the recessive. Since many favourable variations in human society are inherited as dominants whose phenotypic proportions increase as a

result of outbreeding the society encourages exogamous mating. Outbreeding produces bigger and healthier individuals that are more resistant to diseases with better chances of survival. This is called hybrid vigour (Ramalingam, 2000; Verma & Agarwal, 2012)..

2.4.5 Isophenogamy

This type of mating is based on somatic resemblance e.g. body resemblance like the albinos, deaf-mutes etc. When there is mating between individuals of identical or similar phenotypes at a frequency greater than that expected under a system of random mating, the phenomenon is called isophenogamy. The effect of isophenogamy is very similar to that of selfing (Roberts, 1976; Ugwuadu, 2011).

2.4.6 Heterophenogamy

Heterophenogamy refers to mating between non-similar phenotypes to a frequency greater than that which is expected under the system of random mating. If the phenotypes have genetic basis, the effect of heterophenogamy is increase in heterozygosis and accordingly decrease in homozygosis, thus heterophenogamy has similar effect as exogamy or outbreeding (Roberts, 1976; Ugwuadu, 2011). .

2.5 Population Genetics and Health Science

The Hardy-Weinberg formula can be used to calculate the percentage of human population that carries the allele for a particular inherited disease. For example phenylketonuria (PKU) is an inherited inability to breakdown the amino acid phenylalanine. If untreated, the disorder causes severe mental retardation. PKU occurs in about one out of 10,000 babies (1:10,000) born in the United States. New born babies are now routinely tested for PKU and symptoms can be prevented by following strict diet. PKU is due to a recessive allele. Thus, the frequency of individuals in the US population born with PKU corresponds to q2 ratio in the Hardy-Weinberg formula. For one PKU occurrence per 10,000 births, q2 = 0.0001. Therefore, q the frequency of the recessive allele in the population, equals the square root of 0.0001, or 0.01. And P, the frequency of the dominant allele equals 1-q or 0.99 (since p+q = 1). Calculating the frequency of heterozygous individuals who carry PKU allele in a single dosage, the carriers are free from the disorder but may pass the PKU allele on to offspring. Carriers are represented in the Hardy-Weinberg formula by 2pq. That is 2x0.99x0.01 or 0.0198. Thus, the formula tells us that about 2% (actually 1.98%) of US population carries the PKU allele. Estimating the frequency of a harmful allele is essential for any public health programme dealing with genetic diseases (Roberts, 1976; Campbell, Reece &Simon, 2004 & Ugwuadu, 2011).

5. Conclusion

Population genetics has revealed a lot of variations in organisms of the same species through evolution. As a result it has helped to explain how individuals of the same species (plants and animals) tend to become different in structure, anatomy, physiology etc as they evolve over time. Concepts in population genetics like gene flow, genetic drift, isolation, natural selection, Hardy-Weinberg law etc give insights of how evolution occurs when they are disturbed. Socio-culturally, population genetics contributed in understanding mating systems in a population in which exogamy or outbreeding is encouraged while endogamy or inbreeding is discouraged in most cultures. Genetic counseling which is necessary in a population during marriage may not do without some contents of population genetics for proper understanding and application. For example, the Hardy-Weinberg formula can be used to estimate the frequency of a harmful allele which is useful information for public health programme dealing with genetic diseases like PKU, sickle cell anaemia, among others. The concepts of bottle neck effect and founder's effect give insight on the effect of genetic drift on populations all arising from our knowledge of population genetics. Population genetics being a new discipline (Campbell, Reece & Simon, 2004) is worth studying because of its extensive contributions to the existence of living organisms. The students of biological science are exposed to the numerous concepts in this area of biology and their applications which enable them to grasp the topic to ease understanding.

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Revision Questions

1. In MN blood group, the frequency of NN phenotype in American Caucasians is 0.21. Find the frequency of the genotypes of MM, MN and NN individuals using Hardy-Weinberg formula (Fullick, 1994).
2. If p + q = 1 and q2 = 0.36, using the Hardy-Weinberg equation, calculate the frequencies of other genotypes. Comment on your answer.

3(a). In a tabular form, give any four differences between inbreeding and outbreeding.
(b) Briefly explain why society and law do not encourage endogamous marriages among human beings.
4(a) Explain how isolation and natural selection cause evolution in a population.
(b) List four factors that can make a population stay in equilibrium.
5. Write short notes on each of the following terms giving the genetic consequences of each of them.
(a) Gene Pool
(b) Natural Selection
(c) Exogamy
(d) Genetic drift
(e) Gene Flow