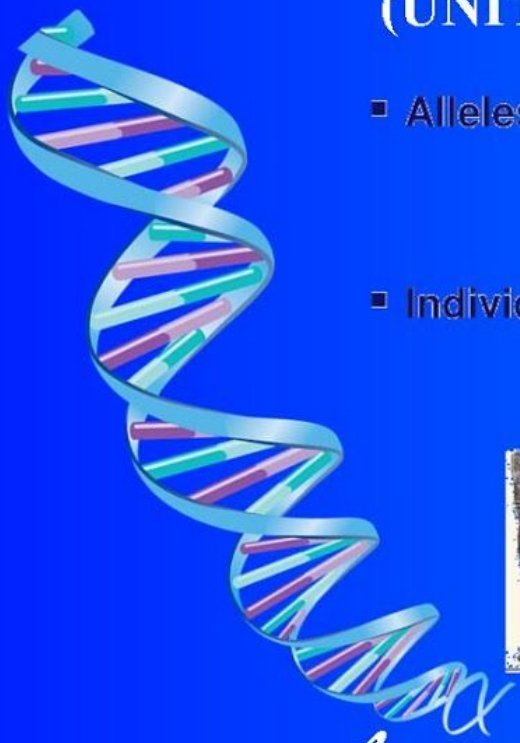
Animal Genetics & Breeding

PRINCIPLES OF ANIMAL AND POPULATION GENETICS

(UNIT - II)



- Alleles: $p + q = 1$

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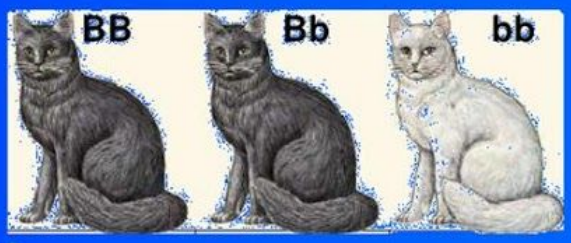

Lecture notes on

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ABOUT

These lecture notes on “Introduction of Population Genetics” were prepared and delivered to my undergraduate students studying Animal Genetics & Breeding course. This course is offered in the second professional year of Bachelor of Veterinary Science & Animal Husbandry degree at College of Veterinary & Animal Sciences, S.V.P.U.A.T, Meerut, Uttar Pradesh, India. This lecture paves the foundation of population genetics. It illuminates students about the intricacies, overview and history of population genetics. This lecture provides the connecting link on how unison of Darwinism and Mendelism gave birth to the field of Population Genetics. The noteworthy contributions of various scientists have been pointed. Use of explanatory boxes and tables have deliberately been used to create an interest among the students. Once through with these lecture notes readers will be able to understand the basics and what they are going to deal in population genetics. I had tried my level best to simplify the concept in easy to understand language. Further constructive suggestions to improve this lecture notes are always welcome from readers on my email and whatsapp.

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1	Probability	20-08-2020	First eprint	Download
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Introduction to Population Genetics

1. Introduction

It has remained one of the intrinsic questions to mankind about how various species on this earth has evolved to its present form. The population of a species have traversed through various variations and keep on changing with time. To understand these things it becomes foremost to study what constitutes a population, its genetic structure and how it gets changed over time. The population genetics is a subfield of genetics that tries to answer these questions. Population genetics have been defined in various ways by different people as given below.

Box 1: Definitions of population genetics

The branch of genetics that deals with the genetic makeup of populations.

The American Heritage® Stedman's Medical Dictionary

The scientific study of the inheritance and prevalence of genes in populations, usually using statistical analysis.

The American Heritage® Science Dictionary

The study of the genetic composition of populations in order to understand the evolutionary forces that select for a particular gene.

The New Dictionary Of Cultural Literacy, Third Edition

Population genetics is a subfield of genetics that deals with genetic differences within and between populations, and is a part of evolutionary biology.

Wikipedia[1]

Population genetics is a field of biology that studies the genetic composition of biological populations, and the changes in genetic composition that result from the operation of various factors, including natural selection.

Okasha, Samir[2]

[1] Retrieved online https://en.wikipedia.org/wiki/Population_genetics

[2] Okasha, Samir, "Population Genetics", The Stanford Encyclopedia of Philosophy (Winter 2016 Edition), Edward N. Zalta (ed.), https://plato.stanford.edu/archives/win2016/entries/population-genetics/

2. History[3]

The history of population genetics can be traversed back to the work of Darwin "Origin of Species", published in 1859. Darwin proposed two theories i) Modern species were descended from common ancestors, and ii) Process of natural selection was the major mechanism of evolutionary change. It is the second theory over which heated debate continued for decades before geneticists could come to a common consensus. Fleeming Jenkins' famously objected to Darwin theory that natural selection to gradually modify a population over a long period of time, in the manner suggested by Darwin, a continual supply of variation is needed. Jenkins argued that given blending inheritance, a sexually reproducing population will become phenotypically homogenous in just a few generations, far shorter than the number of generations needed for natural selection to produce complex adaptations. Darwin was not able to provide a satisfactory answer to objections against his theory. This is because the scientific community at that time was not having a proper understanding of the inheritance mechanism.

Gregor Mendel in 1865 and 1866 proposed principles of inheritance. Obviously, our modern understanding of heredity is vastly more sophisticated than Mendel's, but the key elements of Mendel's theory—discrete hereditary particles that come in different types, dominance and recessiveness, and the law of segregation—have turned out to be essentially correct. Mendel's 'factors' are the genes of modern population genetics, and the alternative forms that a factor can take (e.g. R versus W in the pea plant example) are known as the alleles of a gene. The law of segregation is explained by the fact that during gametogenesis, each gamete (sex cell) receives only one of each chromosome pair from its parent organism. Other aspects of Mendel's theory have been modified in the light of later discoveries. Mendel thought that most phenotypic traits were controlled by a single pair of factors, like seed shape in his pea plants, but it is now known that most traits are affected by many pairs of genes, not just one. Mendel believed that the pairs of factors responsible for different traits (e.g. seed shape and flower colour) segregated independently of each other, but we now know that this need not be so due to linkage.

[3] Excerpted from Okasha, Samir, "Population Genetics", The Stanford Encyclopedia of Philosophy (Winter 2016 Edition), Edward N. Zalta (ed.), https://plato.stanford.edu/archives/win2016/entries/population-genetics/

Despite these points, Mendel's theory marks a turning point in our understanding of inheritance.

The rediscovery of Mendel's work in 1900 did not lead the scientific community to be converted to Mendelism overnight. The dominant approach to the study of heredity at the time was biometry, spearheaded by Karl Pearson in London, which involved statistical analysis of the phenotypic variation found in natural populations. Biometricians were mainly interested in continuously varying traits such as height, rather than the 'discrete' traits such as seed shape that Mendel studied, and were generally believers in Darwinian gradualism. Opposed to the biometricians were the Mendelians, spearheaded by William Bateson, who emphasized discontinuous variation, and believed that major adaptive change could be produced by single mutational steps, rather than by cumulative natural selection à la Darwin. A heated controversy between the biometricians and the Mendelians ensued. As a result, Mendelian inheritance came to be associated with an anti-Darwinian view of evolution.

The Hardy-Weinberg principle, discovered independently by G.H. Hardy and W. Weinberg in 1908, is one of the simplest and most important principles in population genetics (See section 3). The importance of the Hardy-Weinberg principle lies in the fact that it contains the solution to the problem of blending that troubled Darwin. As we saw, Jenkins argued that with sexual reproduction, the variation in the population would be exhausted very rapidly. But the Hardy-Weinberg principle teaches us that this is not so. Another benefit of the Hardy-Weinberg principle is that it greatly simplifies the task of modelling evolutionary change. When a population is in Hardy-Weinberg equilibrium, it is possible to track the genotypic composition of the population by directly tracking the allelic frequencies (or gametic frequencies).

Population genetics as we know it today arose from the need to reconcile Mendel with Darwin, a need which became increasingly urgent as the empirical evidence for Mendelian inheritance began to pile up. The first significant milestone was R.A. Fisher's 1918 paper, 'The Correlation between Relatives on the Supposition of Mendelian Inheritance', which showed how the biometrical and Mendelian research traditions could be unified. Fisher demonstrated that if a given continuous trait, e.g. height, was affected by a large number of Mendelian factors, each of which made a small

difference to the trait, then the trait would show an approximately normal distribution in a population. Since the Darwinian process was widely believed to work best on continuously varying traits, showing that the distribution of such traits was compatible with Mendelism was an important step towards reconciling Darwin with Mendel.

The full reconciliation was achieved in the 1920s and early 30s, thanks to the mathematical work of Fisher, Haldane and Wright. Each of these theorists developed formal models to explore how natural selection, and other evolutionary forces such as mutation, would modify the genetic composition of a Mendelian population over time. This work marked a major step forward in our understanding of evolution, for it enabled the consequences of various evolutionary hypotheses to be explored quantitatively rather than just qualitatively. Verbal arguments about what natural selection could or could not achieve, or about the patterns of genetic variation to which it could give rise, were replaced with explicit mathematical arguments. The strategy of devising formal models to shed light on the process of evolution is still the dominant research methodology in contemporary population genetics.

There were important intellectual differences between Fisher, Haldane and Wright, some of which have left legacies on the subsequent development of the subject. One difference concerned their respective attitudes towards natural selection. Fisher and Haldane were both strong Darwinians—they believed that natural selection was by far the most important factor affecting a population's genetic composition. Wright did not downplay the role of natural selection, but he believed that chance factors (Random drift) also played a crucial role in evolution, as did migration between the constituent populations of a species. A related difference is that Wright emphasized epistasis, or non-additive interactions between the genes within a single genome, to a much greater extent than Fisher or Haldane. Despite these differences, the work of all three was remarkably consonant in overall approach.

Population genetics came into being in the 1920s and 1930s, long before the molecular structure of genes had been discovered. In these pre-molecular days, the gene was a theoretical entity, postulated in order to explain observed patterns of inheritance in breeding experiments; what genes were made of, how they caused phenotypic changes, and how they were transmitted from parent to offspring were not known. Even without knowing what these hereditary particles are made of, or how they exert their

phenotypic effects, the early population geneticists were able to devise an impressive body of theory. That the theory continues to be useful today illustrates the power of abstract models in science. Today we do know the answers to these questions, thanks to the spectacular success of the molecular genetics ushered in by Watson and Crick's discovery of the structure of DNA in 1953. The gene has gone from being a theoretical entity to being something that can actually be manipulated in the laboratory.

Modern population genetics has built on this theoretical edifice in a number of ways, most notably by integrating the theories of classical population genetics with data from molecular biology. More recently, extensive data sets on variation at the DNA rather than the protein level have become available; this has led to the rise of 'molecular population genetics' and an associated set of ideas known as coalescent theory[4]. Unlike traditional population-genetic analysis, which tries to determine how a given population will evolve in the future, coalescent theory tries to reconstruct the ancestral state of a population from its current state, based on the idea that all the genes in a population ultimately derive from a single common ancestor. Coalescent theory underpins much contemporary research in population genetics.

3. Overview

One of the essential building blocks for the subject of population genetics is population. A population is composed of members of the same species that simultaneously live and interact in the same area. When individuals in a population breed, they pass down their genes to their offspring. Many of these genes are polymorphic, meaning that they occur in multiple variants. Such variations of a gene are referred to as alleles. The collective set of all the alleles within a population is known as the gene pool. The term gene pool can be defined as the sum of a population's genetic material at a given time. The term typically is used in reference to a population made up of individuals of the same species and includes all genes and combinations of genes (sum of the alleles) in the population.[5]

While some alleles of a given gene might be observed commonly, other variants may be encountered at a much lower frequency. Population genetics examines genetic variation

[4] Wakeley, J., 2004, Coalescent Theory: an Introduction, Greenwood Village, CO: Roberts.
[5] Retrieved online https://www.britannica.com/science/gene-pool

within and between populations, and changes in allele frequencies across generations. Population geneticists use mathematical models to investigate and predict allele frequencies in populations. These allele frequencies across generations can be explained with the help of Hardy - Weinberg law. This law infers that allele and genotype frequencies in a population will remain constant from generation to generation in the absence of other evolutionary influences. These influences include genetic drift, mate choice, assortative mating, natural selection, sexual selection, mutation, gene flow, meiotic drive, genetic hitchhiking, population bottleneck, founder effect and inbreeding.

Table 1: Short description of various evolutionary influences acting over population

Evolutionary Influence	Description
Genetic drift[6]	Genetic drift, also called genetic sampling error or Sewall Wright effect, a change in the gene pool of a small population that takes place strictly by chance. Genetic drift can result in genetic traits being lost from a population or becoming widespread in a population without respect to the survival or reproductive value of the alleles involved. A random statistical effect, genetic drift can occur only in small, isolated populations in which the gene pool is small enough that chance events can change its makeup substantially. In larger populations, any specific allele is carried by so many individuals that it is almost certain to be transmitted by some of them unless it is biologically unfavourable.
Mate choice[7]	Mate choice can be simply defined as any pattern of behavior shown by members of one sex that leads to preferred mating with certain members of the opposite sex.
Assortative mating[8]	Assortative mating is the tendency for people to choose mates who are more similar (positive) or dissimilar (negative) to themselves in phenotype characteristics than would be expected by chance. If these characteristics are genetically determined, positive assortative mating may increase homozygosity in the population. An important

[6] Retrieved online https://www.britannica.com/science/genetic-drift
[7] Leslie A. Knapp, in Encyclopedia of Reproduction (Second Edition), 2018
[8] H. Richard Johnston, ... Stephanie L. Sherman, in Emery and Rimoin's Principles and Practice of Medical Genetics and Genomics (Seventh Edition), 2019

	difference between inbreeding and positive assortative mating is that inbreeding affects all loci, while assortative mating affects only those that play a role in the phenotype characteristics that are similar.
Natural selection[9]	Natural selection is the differential survival and reproduction of individuals due to differences in phenotype. It is a key mechanism of evolution, the change in the heritable traits characteristic of a population over generations. Charles Darwin popularised the term "natural selection", contrasting it with artificial selection, which in his view is intentional, whereas natural selection is not.
Sexual selection[10]	Sexual selection is a mode of natural selection in which members of one biological sex choose mates of the other sex to mate with (intersexual selection), and compete with members of the same sex for access to members of the opposite sex (intrasexual selection).
Mutation[11]	Mutations are essential to evolution. Every genetic feature in every organism was, initially, the result of a mutation. The new genetic variant (allele) spreads via reproduction, and differential reproduction is a defining aspect of evolution. It is easy to understand how a mutation that allows an organism to feed, grow or reproduce more effectively could cause the mutant allele to become more abundant over time. Soon the population may be quite ecologically and/or physiologically different from the original population that lacked the adaptation. Even deleterious mutations can cause evolutionary change, especially in small populations, by removing individuals that might be carrying adaptive alleles in other genes.
Gene flow[12]	Gene flow is also called gene migration. Gene flow is the transfer of genetic material from one population to another. Gene flow can take place between two populations of the same species through migration, and is mediated by reproduction and vertical gene transfer from parent to offspring. Alternatively, gene flow can take place between two different species through horizontal gene transfer (HGT, also known as lateral gene transfer), such as gene transfer from bacteria or viruses to a higher organism, or gene transfer from an endosymbiont to the host.

[9] Retrieved online https://en.wikipedia.org/wiki/Natural_selection
[10] Retrieved online https://en.wikipedia.org/wiki/Sexual_selection
[11] Carlin, J. L. (2011) Mutations Are the Raw Materials of Evolution. Nature Education Knowledge 3(10):10
[12] Supratim Choudhuri, in Bioinformatics for Beginners, 2014

Meiotic drive[13]	Meiotic drive is a natural phenomenon that favors the non-Mendelian transmission of an allele to progeny to achieve higher than expected allele frequencies.
Genetic hitchhiking[14]	When a favourable mutation arises, and increases to fixation, it gives a fortuitous advantage to all the genes with which it was originally associated. Maynard Smith & Haigh (1974) termed this process `hitchhiking' and showed that, in large populations, it could reduce neutral diversity much more than random genetic drift. It may be considered as indirect effects of selection at one or more loci on the rest of the genome. Selection may be for alleles which are unconditionally favourable; it may act to eliminate deleterious mutations; or it may fluctuate in space or time. The surrounding loci may be neutral; they may modify the genetic system; or they may themselves be directly selected. The same processes underlie all these various cases, and so it is illuminating to consider them together. Indeed, the idea of `hitchhiking' brings together apparently diverse aspects of population genetics.
Population bottleneck[15]	The bottleneck effect refers to the way in which a reduction and subsequent increase in a population's size affects the distribution of genetic variation among its individuals. Typically, a population bottleneck reduces genetic variance at genetic loci and increases nonrandom associations between different loci as a result of the increased importance of genetic drift. Analysis of multilocus genetic data using a range of different genetic statistics can allow inferences to be made about the timing and strength of a population bottleneck in the past. Population bottlenecks are thought to be responsible for the very low levels of genetic variation found in a number of species that now have large population sizes, including humans and the laboratory model Drosophila melanogaster.
Founder effect[16]	The term founder effect, proposed by Mayr, describes the establishment of a new population by a few original founders, which carries only a small fraction of the total genetic variation of the original population. A founder effect in a population is suspected when there is an unusually high prevalence of some genetic disorder and/or a very

[13] James K. Biedler, ... Zhijian J. Tu, in Genetic Control of Malaria and Dengue, 2016

[14] Barton, N. H. "Genetic Hitchhiking." Philosophical Transactions: Biological Sciences, vol. 355, no. 1403, 2000, pp. 1553–1562. JSTOR, www.jstor.org/stable/3066883.

[15] J.R. Pannell, in Brenner's Encyclopedia of Genetics (Second Edition), 2013

[16] Gabriela Chavarriá-Soley, ... Henriette Raventoś, in Genome Plasticity in Health and Disease, 2020

	low prevalence of others.
Inbreeding[17,18]	Inbreeding is defined as the probability of two alleles in an individual being identical by descent, and is normally the result of mating related individuals. Inbreeding results in reductions in fitness (inbreeding depression) in all naturally outbreeding species, and to a lesser extent in species with inbreeding mating systems. All fitness traits are susceptible to inbreeding depression, but traits peripheral to fitness exhibit little or no inbreeding depression.

There is an alternative body of theory, known as quantitative genetics, which deals with so-called 'polygenic' or 'continuous' traits, such as height, which are thought to be affected by genes at many different loci in the genome, rather than just one or two[19]. Quantitative genetics employs a quite different methodology from population genetics. The latter, as we have seen, aims to track gene and genotype frequencies across generations. By contrast, quantitative genetics does not directly deal with gene frequencies; the aim is to track the phenotype distribution, or moments of the distribution such as the mean or the variance, across generations. Though widely used by animal and plant breeders, quantitative genetics is usually regarded as a less fundamental body of theory than population genetics, given its 'phenotypic' orientation, and plays less of a role in evolutionary theorising. Nonetheless, the relationship between population and quantitative genetics is essentially harmonious.

4. Summary

We have learnt in this lecture that principles of population genetics attempt to explain the genetic diversity in present populations and the changes in allele and genotype frequencies over time. There was a long ensued scientific ideology of Mendelism and Darwinism that prevailed over decades to prove which one is a better and correct explanation to evolutionary biology. However, with time and various landmark discoveries by scientific community unison between two ideologies occurred that fine tuned our basic knowledge about population genetics. Population geneticists use

[17] James M. Myers, ... Robert N. Iwamoto, in Reproductive Biotechnology in Finfish Aquaculture, 2001
[18] Richard Frankham, in Encyclopedia of Ecology (Second Edition), 2019
[19] Falconer, D.S., 1995, Introduction to Quantitative Genetics, 4th edition, London: Longman.

mathematical models to investigate and predict allele frequencies in populations. These allele frequencies across generations can be explained with the help of Hardy - Weinberg law. This law infers that allele and genotype frequencies in a population will remain constant from generation to generation in the absence of other evolutionary influences. A brief introduction about these influences has been covered that includes genetic drift, mate choice, assortative mating, natural selection, sexual selection, mutation, gene flow, meiotic drive, genetic hitchhiking, population bottleneck, founder effect and inbreeding.

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ABOUT THE AUTHOR

Dr. Kuldeep Kumar Tyagi had completed his B.V.Sc & A.H. in the year 2006 from Guru Angad Dev Veterinary and Animal Sciences University, Ludhiana, Punjab India. He got admission in a master program in the subject of Animal Genetics and Breeding at Indian Veterinary Research Institute, Bareilly, Uttar Pradesh, India after securing 6th rank in All India ICAR-JRF examination. He had completed his Masters in the year 2008 and carried out research on competent fibroblast cells used in somatic cell nuclear transfer. He qualified CSIR Net in his first attempt during the final semester of masters program itself. He got selected as Assistant Professor in the year 2009 at College of Veterinary Science & A.H. at Navsari Agricultural University, Navsari, Gujarat, India. He enriched his practical knowledge and expertise in the subject of Animal Breeding while disbursing his duties as Scheme Incharge at Livestock Research Station of the same university for 9 years. During the same tenure he also accumulated practical expertise on various aspects of field level breeding programs while heading "All India Coordinated Research Project on Goat Improvement - Surti Field Unit" as Principal Investigator. He completed his Ph.D. in the year 2016 from the same university as an inservice candidate. He had worked on gene expression studies on mammary epithelial cells of buffaloes during his Ph.D. degree program. He had been selected as Associate Professor in the department of Animal Genetics & Breeding, College of Veterinary and Animal Science, Sardar Vallbhbhai, Patel University of Agriculture & Technology, Meerut, Uttar Pradesh, India in the year 2018. Since then he has been heading the same department as Officer-Incharge. He had handled 5 externally funded and 27 institutionally funded research projects. He had coguided two masters students. He has in his credit 61 research papers, 14 research recommendations, 4 lecture notes and 4 success stories. He is a member of 4 professional societies and attended 21 conferences/ symposiums/ workshops. He has remained on a panel of experts for framing question papers for various Universities and National level examination bodies. He is hosting a google site for online teaching https://sites.google.com/view/learnagb and can be reached at drtyagivet@gmail.com for initiating a conversation.

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