Chapter.2. Genetic of population

Introduction

A population is a group of individuals of the same species, each having a certain chance of being mated with another individual. The concept of population thus involves spatial, temporal, and genetic criteria and results from the fact that individuals of the same species do not all have the opportunity to meet and interbreed due to geographical distance and habitat heterogeneity.

The objective of population genetics is the study of gene and genotype frequencies, and the factors likely to modify these frequencies over successive generations. Some of these factors, such as selection, mutations, genetic drift, and migrations, can change gene and genotype frequencies.

Consanguinity (mating between related individuals) can modify genotype frequencies without influencing gene frequencies.

1. Genetic Constitution of a Population: Gene and Genotype Frequencies

The genetic structure of a population is both its genotypic and allelic structure.

1.1. Genotypic Frequency

Consider a population in which a trait is determined by a locus with two alleles, B and b. The possible genotypes are BB, Bb, and bb. Suppose that among the n individuals in the population, n_1 individuals have genotype BB, n_2 have genotype Bb, and n_3 have genotype bb.

$(n_1 + n_2 + n_3 = n)$

The genotypic frequency of a particular genotype is equal to the number of individuals with that genotype divided by the total number of individuals in the population. Thus:

$$F(BB) = n_1 / n$$

$$F(Bb) = n_2 / n$$

$$F(bb) = n_3 / n$$

Note that the sum of genotypic frequencies in a population equals 1:

$$F(BB) + F(Bb) + F(bb) = 1$$

1.2. Allelic or Gene Frequency

The frequency of an allele in a population of n individuals is equal to the number of alleles of that type divided by the total number of alleles at that locus in the population. Since there are two alleles at a given locus, the total number of alleles in the population is 2n.

However, individuals with genotype BB possess two B alleles at the locus, while individuals with genotype Bb possess only one.

The allelic frequency of B is:

$$f(B) = (2n_1 + n_2) / 2n = n_1/n + ½\, n_2/n = F(BB) + ½\, F(Bb)$$

Similarly, the frequency of b is:

$$f(b) = (2n_3 + n_2) / 2n = n_3/n + \frac{1}{2} n_2/n = F(bb) + \frac{1}{2} F(Bb)$$

It is noted that the sum of allelic frequencies equals 1:

$$f(B) + f(b) = 1$$

These allelic frequencies are generally denoted as:

$$f(B) = p \text{ and } f(b) = q$$

Allelic frequencies for a particular locus in a group of individuals can be determined from knowledge of genotype frequencies. Let's take an example. Suppose there are two alleles A and a. Let's now classify 100 individuals and count the individuals belonging to each genotype:

Genotype	Number
AA	30
Aa	60
aa	10
Total	100

Each individual possesses two genes (diploid species), so there are 200 genes present at this locus. Each AA individual possesses two A genes, and each Aa individual possesses one; thus, in the example, there are 120 A genes and 80 a genes.

The frequency of A is therefore 60% or 0.6 (120/200), and that of a is 40% or 0.4 (80/200). To express this relationship more generally, let's represent the frequencies of genes and genotypes as follows:

Let:

$$R = F(AA)$$

$$S = F(Aa)$$

$$T = F(aa)$$

Thus:

$$p = f(A),\ q = f(a)$$

So that:

$$p + q = 1 \text{ and } R + S + T = 1$$

Since each individual possesses two genes, the frequency of gene A is $\frac{1}{2}(2R + S)$, and the relationship between gene frequencies and genotype frequencies among the counted individuals is:

$$p = R + \frac{1}{2} S$$

$$q = T + \frac{1}{2} S$$

When the genotype-phenotype relationship is direct:

This is the case of codominance where number of genotypes = number of phenotypes.

Example: 2 alleles A and B.

AA [A] with count n_1

Aa [Aa] with count n_2

aa [a] with count n_3

When the genotype cannot be directly deduced from the phenotype:

This is the case of dominance: the genotype cannot be deduced from the phenotype (number of genotypes ≠ number of phenotypes), so the calculation of allelic frequencies is not directly possible. Estimating gene frequencies from genotypes is only possible if all genotypes are identifiable: the two alleles are codominant. To calculate allelic frequencies in a case of dominance, one must assume the population is in Hardy-Weinberg equilibrium to estimate the allelic frequencies.

2. Hardy-Weinberg Equilibrium

The Hardy-Weinberg (HW) law was proposed independently in 1908 by the English mathematician Hardy and the German physician Weinberg.

The Hardy-Weinberg law is defined as follows:

In an infinitely large population, where mating is random (panmixia), where there is no migration, no selection against a particular phenotype, and where the mutation rate is constant or mutations are absent, the proportions of different genotypes remain constant from one generation to the next.

2.1. Conditions for Hardy-Weinberg Equilibrium

Five (5) conditions are therefore necessary for Hardy-Weinberg equilibrium:

1. Large population size.
2. Random mating (panmixia).
3. No migration (no movement of individuals between populations).
4. No mutation (no appearance of new genes or disappearance of existing ones).
5. No selection (all gametes, regardless of type, have an equal probability of contributing to the next generation).

2.2. Demonstration of the H-W Law

Consider an autosomal locus with 2 alleles A and a at generation n. The frequencies of genotypes AA, Aa, and aa are respectively R, S, and T, while the frequencies of genes A and a are respectively p and q.

The genotypic structure of generation n+1 results from the random meeting of male and female gametes.

At generation n, the frequencies of gametes carrying A is p, and those carrying a is q.

Random union of gametes (Panmixia) gives:

	Male Gametes	
Female Gametes	A (p)	a (q)
A (p)	AA (p^2)	Aa (pq)
a (q)	Aa (pq)	aa (q^2)

Thus, in generation n+1:

1. Frequency of AA = p^2
2. Frequency of Aa = 2pq
3. Frequency of aa = q^2

The sum is $p^2 + 2pq + q^2 = (p + q)^2 = 1$.

At generation n+1, the frequency of A (p') is:

$p' = F(AA) + \frac{1}{2} F(Aa) = p^2 + \frac{1}{2} (2pq) = p^2 + pq = p(p + q) = p$

Similarly, $q' = q$.

The allelic frequencies remain unchanged. Therefore, under the assumed conditions, the genotypic frequencies will be p^2, $2pq$, q^2 at generation n+1 and will remain so for subsequent generations. This is the Hardy-Weinberg equilibrium.

3. Heritability

In a population, for a given trait, the variance of performance, or phenotypic variability, has a dual origin: heredity (more specifically, the additive effects of genes), leading to genetic variability (additive), and the environment, leading to environmental variability.

Phenotypic variability = Genetic variability + Environmental variability.

Only genetic variability is of interest to the breeder; environmental variability, or differences between individuals arising from environmental effects, is not heritable.

The effectiveness of selection is linked to the existence of genetic variability. It is therefore logical to define a coefficient indicating what proportion of the phenotypic variability is of genetic origin; this is the heritability coefficient, denoted h^2. In this case, individual selection is ineffective. Family selection (based on the performance of relatives) or progeny testing (which provides information on the breeding value of a male based on the performance of his offspring) is preferred, as it becomes more effective.

3.1. Heritability in Animal Breeding

In animal breeding, the concept of heritability takes on a crucial practical and economic dimension. It is no longer just about understanding the sources of variation, but about predicting and optimizing genetic progress within a herd or breed.

Here is how heritability is concretely used in livestock farming.

3.1.1. Definition in a Breeding Context

In animal breeding, heritability (still denoted as h^2) retains the same statistical definition: it is the proportion of the difference between animals (phenotypic variance) that is due to additive genetic effects.

Why are additive effects so important here?

Because these are the only genetic effects that are reliably passed from parents to offspring. If you select a bull with very good additive effects for milk production, you have a good chance that his daughters will also have good production.

3.1.2. Practical Utility: Selecting Efficiently

Heritability is the tool that answers the fundamental question for breeders or selectionists: "If I choose this particular breeding animal, how quickly will I improve my herd?"

Breeding traits are generally classified into three categories based on their heritability:

A. High Heritability Traits ($h^2 > 0.30 – 0.40$)

Examples:

- Butterfat content (milk fat), protein content, weight at a given age, height at withers.

Implication in breeding: Individual selection (choosing the best animals based on their own performance) is highly effective. Genetic progress is rapid. If you want to improve milk protein percentage, you simply select cows with naturally high levels and breed them.

B. Moderate Heritability Traits (h^2 between 0.15 and 0.30)

Examples: Milk yield, average daily gain (ADG), feed efficiency.

Implication in breeding: Selection works, but more precise methods are needed, such as progeny testing (evaluating a male based on the performance of his daughters) to be sure of the genetic value.

C. Low Heritability Traits ($h^2 < 0.10 – 0.15$)

Examples: Fertility (success rate of insemination), litter size (number of offspring per litter), longevity, disease resistance, mortality.

Implication in breeding: Individual selection is very slow and ineffective because the environment (care, nutrition, stress) completely masks the genetic potential. For these traits, the focus is on:

1. Improving rearing conditions (this is the fastest lever).
2. Genomic selection (DNA analysis) to identify favorable genes.
3. Crossbreeding (to benefit from heterosis or hybrid vigor).

3.1.3. The Link with Breeding Value (Index)

In animal breeding, heritability is not used alone. It is used to calculate the animal's Breeding Value (BV), often called an index.

The simplified formula is: $BV = h^2 \times (P - M)$

· P is the animal's performance (e.g., 10,000 liters of milk).

· M is the herd or breed average (e.g., 8,000 liters).

· h^2 is the heritability of the trait.

If milk production has a heritability of 0.25 (low to moderate), and a cow produces 2,000 liters above average, her estimated "breeding value" will only be 0.25 \times 2000 = 500 liters.

This means: Only 500 liters of her advantage are estimated to be due to her genes (and therefore transmissible); the remaining 1,500 liters are due to good nutrition, lack of illness, etc. (the environment).

3.1.4. The Impact of Environment and Interactions

Heritability in animal breeding is not fixed. It depends on the context:

- Level of environmental homogeneity: On an experimental farm where all animals are fed the same way and housed under the same conditions, the apparent heritability of production traits will be higher (because environmental differences have been eliminated). In real-world conditions, across thousands of different farms, the calculated heritability will be lower because the environment adds variation.
- Genotype x Environment interaction: Sometimes, the "best" bull in an intensive farming system (using grains) is not the best in an extensive system (on pasture). This means that gene expression (and thus heritability) depends on the environment.

3.1.5. Why Is This Crucial Today?

With the advent of genomic selection (DNA analysis), it is now possible to estimate an animal's breeding value from birth without waiting for its performance.

- For a trait with high heritability (e.g., meat quality), genomics merely confirms what was already observable.
- For a trait with low heritability (e.g., fertility), genomics is a revolution. It makes it possible to identify breeding animals carrying the "good versions" of genes, something

that was previously almost impossible because such effects were always confounded with environmental effects.

Trait Type Heritability (h^2) Examples Selection Method Speed of Progress:

- Physical / Production High (> 0.3) Fat content, weight, height Individual performance Fast
- Growth Moderate (0.15–0.3) Weight gain, milk yield Progeny testing / Genomics Moderate
- Functional / Health Low (< 0.15) Fertility, survival, resistance Genomics / Environmental management Slow

In short, in animal breeding, heritability is the compass of genetic progress: it indicates to breeders and technicians which levers (genetics or environment) to apply to improve their livestock most effectively.

References

Bousbia, A (2015): Sélection et Amélioration génétique: génétique qualitative, génétique des populations, génétique statistique et génétique quantitative. Polycopié pour la licence production animale. Département d'écologie et génie de l'environnement, Faculté des sciences de la nature et de la vie et des sciences de la terre et de l'univers, Université 8 Mai 1945, Guelma.

Falconner, D.S et Mackay, T.F.C (1996) : Introduction to quantitative genetics, *4th ed. Longman*.