Chapter 1 : Qualitative genetics and its applications in agronomic sciences

Introduction

Genetics is the science of heredity. It studies how genetic material is organized at the cellular level, how it is transmitted from parents to offspring, and how it is expressed in the traits of an individual. In animal production, genetics provides the tools to understand why animals differ for important traits such as growth, milk yield, fertility, coat color or presence of horns, and how these differences can be used in breeding programs.

Within genetics, qualitative genetics focuses on traits controlled by one or a few major genes, which follow Mendelian inheritance and show discrete phenotypic categories. Examples include horned vs polled, black vs red coat, or the presence vs absence of a simple genetic defect.
Understanding qualitative genetics and gene interactions is a first step before studying quantitative traits and complex selection schemes in livestock.

Section 1 – Interactions Between Allelic and Non-allelic Genes

I. Qualitative genetics: definition and main features

I.1. Definition of qualitative genetics

Qualitative genetics studies the expression and transmission of genes controlling Mendelian or qualitative traits, where phenotypes can be clearly divided into distinct classes (for example, presence or absence, one color vs another).
These traits are usually determined by one or a small number of loci, each with a limited series of alleles, often only two alleles per locus, and they follow simple Mendelian segregation in families.

I.2. Characteristic features of qualitative traits

Qualitative traits show several characteristic features:

1. ***Simple genetic determinism***
   A qualitative trait is controlled by one or a few loci, each with a small number of alleles. The effect of each gene is large and strongly influences the phenotype.

2. ***Major-gene effect***
   Because each gene has a major effect, the genotype at the main locus can often be deduced from the visible phenotype. For example, an obviously horned buck is very likely to be homozygous for the horn allele at the horn locus.

3. ***Discontinuous phenotypic distribution***
   Phenotypes fall into discrete categories (horned vs polled, black vs red, normal vs affected) rather than forming a continuous gradient, which is typical for quantitative traits.

4. ***Limited environmental influence***
   For qualitative traits, the environment usually has little influence on whether the gene is expressed; the phenotype reflects mainly the genotype, with minimal modulation by external conditions.
   Horn status in goats is a classical qualitative trait: animals are either horned or polled, and this difference is largely determined by a single major locus.

II. Expression and interactions of allelic genes

II.1. Allelic genes: basic concepts

At a given locus, different versions of the same gene are called alleles, and they occupy the same position on homologous chromosomes.
In a diploid animal, each locus is present in two copies, so each individual carries two alleles at each locus. If the two alleles are identical, the individual is homozygous at that locus; if they are different, the individual is heterozygous.

Example (horn locus in goats):

- At the horn locus, we can distinguish allele P (polled, absence of horns) and allele p (horned, presence of horns).
- Possible genotypes at this locus are P//P, P//p, and p//p, where "//" represents the pair of homologous chromosomes.

The genotype is the allelic combination at a locus (for example p//p), whereas the phenotype is the observable trait (for example horned).

II.1.1. Complete dominance

a) General definition

Complete dominance describes the situation where one allele (dominant) completely masks the phenotypic effect of the other allele (recessive) in heterozygous individuals.
For a locus with alleles A (dominant) and a (recessive), three genotypes are possible (A//A, A//a, a//a), but only two phenotypes are observed: [A] (dominant phenotype) and [a] (recessive phenotype).

In such a system:

- The heterozygote A//a has the same phenotype as the homozygous dominant A//A.

- The recessive phenotype [a] appears only in homozygous recessive animals a//a.

b) Biological basis of dominance

Several mechanisms can explain dominance:

- The recessive allele may be a loss-of-function mutation producing no functional protein, while the wild-type allele produces a normal protein responsible for the phenotype.
- For many genes, the amount of protein produced by one functional allele is sufficient to produce the full trait, so the heterozygote shows the same phenotype as the homozygous dominant.

Animal illustration:

1. ***Polled vs horned in cattle and goats***
   In several breeds, a dominant allele at the horn locus leads to polled animals. Horned animals are usually homozygous for the recessive horn allele. This trait is used in breeding to reduce dehorning and improve animal welfare.

Figure 1 : Variation in horn phenotype as a qualitative trait in sheep (horned and polled) (Johnston et al., 2010)

2. ***Diluted coat color in Charolais cattle***
   In the Charolais breed, a specific dominant allele at a coat-color locus produces a characteristic diluted white or cream color. This color is often used as a visual signal of Charolais origin in crossbred animals.
3. ***Skin color in chickens***
   Alleles at a skin-color locus in chickens determine whether the skin appears white or yellow. Only birds with two copies of the recessive allele can deposit carotenoid pigments from feed and therefore show yellow skin, whereas birds with the dominant allele remain white.

II.1.2. Codominance

a) General definition

Codominance is an allelic interaction in which two different alleles are both fully expressed in the heterozygous phenotype.
In this case, the heterozygote is not intermediate; instead, both allelic products can be detected simultaneously, and the three genotypes correspond to three distinguishable phenotypes.

For alleles A and B:

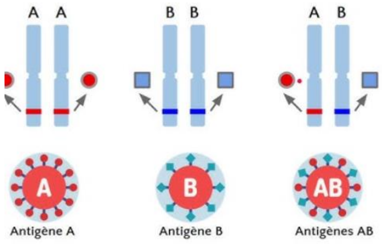
- Genotypes: A//A, A//B, B//B.
- Phenotypes: [A], [A and B], [B].

b) Difference from incomplete dominance

- In codominance, both alleles are expressed; the phenotype of the heterozygote shows features of both homozygotes.
- In incomplete dominance, the heterozygote shows a reduced or intermediate expression of a single active allele, and the phenotype lies between the two homozygotes.

Animal illustration

1. ***Blood group systems***
   In many mammals, blood group antigens are encoded by codominant alleles. Heterozygous animals express both forms of antigen on the surface of their red blood cells, which can be detected serologically.



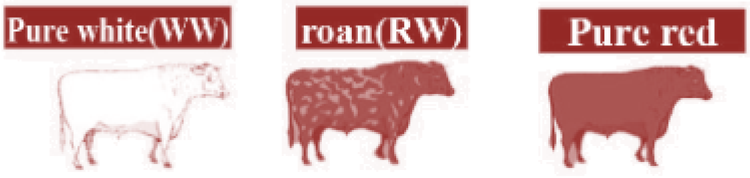
2. ***Kappa-casein in dairy cattle***
   At the kappa-casein locus in cattle, there are at least two common alleles, A and B, which differ in their effects on cheese yield and curd firmness.
    - Genotypes: A//A, A//B, B//B.
    - Phenotypes: [A], [A and B], [B], because both A and B protein variants are present in the milk of heterozygous cows.

This codominance is exploited in dairy breeding, as the B allele is often associated with better cheese-making properties and is therefore favored in certain breeds.

II.1.3. Incomplete dominance

Definition

Incomplete dominance is an allelic interaction in which the phenotype of the heterozygote is intermediate between those of the two homozygotes.
All three genotypes (for example R//R, R//r, r//r) give three distinct phenotypes, and the F2 generation from a monohybrid cross shows a 1:2:1 ratio for both genotypes and phenotypes.



Animal illustration

At the coat color locus in Shorthorn cattle, two alleles exist: R (red) and r (white).

- RR: uniformly red coat.
- Rr: roan coat (intermediate: intimate mix of red and white hairs).
- rr: uniformly white coat.
  Unlike codominance, both alleles contribute to a **blended** phenotype, not side-by-side expression.

Cross of pure red line (RR) × pure white line (rr):

All F1 are heterozygotes Rr → roan coat (intermediate phenotype).

Selfing of F1 (Rr × Rr):

Gametes	**R (red)**	**r (white)**
R	RR (red)	Rr (roan)
r	Rr (roan)	rr (white)

F2 results: 25% RR (red), 50% Rr (roan), 25% rr (white).

Phenotypic ratio 1:2:1, matching the genotypic ratio—hallmark of incomplete dominance.

II.1.4. Incomplete penetrance

Definition

Penetrance is the proportion of individuals with a given genotype that actually express the expected phenotype.
When all individuals with the genotype show the trait, penetrance is complete (100%). When only some individuals show the trait, penetrance is incomplete (for example 90%).

Penetrance is usually expressed as a fraction or percentage for a genotype in a specific population and environment.

Causes of incomplete penetrance

Incomplete penetrance can result from:

- Environmental conditions that modify gene expression or development.
- Modifier genes that enhance or reduce the effect of the main gene.
- Epigenetic mechanisms such as DNA methylation or non-coding RNAs influencing gene activity.

Animal illustration (double-muscled cattle, conceptual)

A gene responsible for muscular hypertrophy ("double muscling") in cattle may show incomplete penetrance: not all animals with the genotype associated with double muscling present the typical phenotype.
If 90% of animals with genotype mh//mh display the double-muscled appearance and 10% look normal, the penetrance of the mh allele is 0.9 (90%) under the considered conditions.

II.1.5. Variable expressivity

Definition

Expressivity refers to the degree or intensity with which a given genotype is expressed in different individuals.
When expressivity is variable, all individuals share the same genotype, but the trait appears more or less strongly expressed from one individual to another.

Penetrance answers "Is the trait present, yes or no?", whereas expressivity answers "To what extent is the trait expressed?".

Animal illustration (double-muscled phenotype)

Among double-muscled cattle with genotype mh//mh, some animals show very pronounced muscle development, extreme conformation and associated problems, while others display a milder muscular

hypertrophy.
This variability in intensity, despite the same genotype, illustrates variable expressivity of the double-muscled gene.

II.1.6. Allelic series and multiple alleles

Definition

A series of alleles at a locus is a set of more than two alternative alleles that can occupy the same chromosomal site in a population.
Although each individual carries only two alleles at a time (one on each homologous chromosome), multiple alleles in the population allow a wide range of genotypes and phenotypes.

These alleles may be ranked according to their effect on a trait, forming a functional series from “strong” to “weak” alleles, or they may show different dominance relationships.

Animal illustration (casein alleles in goats)

At the alpha-s1 casein locus in goats, numerous alleles have been described.
A subset of these alleles is exploited in selection because they differ in their effect on milk protein content: some are associated with a high protein yield, others with intermediate values, and others with low protein content.

This allelic series provides breeders with a tool to modify milk composition by choosing appropriate genotypes for breeding.

II.2. Non-allelic genes and their interactions

Non-allelic or intergenic interactions occur when different loci influence the same trait and interact in their expression.
Important types of non-allelic interactions in qualitative genetics include pleiotropy, epistasis, sex-linked inheritance, and lethal or undesirable alleles.

II.2.1. Pleiotropy

Definition

Pleiotropy occurs when a single gene influences two or more distinct and seemingly unrelated traits.
This happens when the gene product (protein) participates in several physiological processes or developmental pathways, so that one genetic change has multiple phenotypic consequences.

Consequences for breeding

Pleiotropic genes can create conflicts in breeding decisions: improving one trait through selection on a gene may cause undesired changes in another trait controlled by the same gene.
This is particularly important when production-enhancing alleles also have negative effects on fertility, health or animal welfare.

Animal illustration (polled gene and fertility in goats)

In goats, the polled allele P (absence of horns) has a positive effect on horn status, but in the homozygous state (P//P) it can be associated with reproductive disorders and infertility in females.
The same gene thus affects horn status and fertility, illustrating pleiotropy and forcing breeders to manage polled genotypes carefully.

II.2.2. Epistasis

Definition

Epistasis is a type of non-allelic interaction in which one gene (epistatic gene) masks or modifies the phenotypic expression of another gene (hypostatic gene) that acts on the same trait.
In dihybrid crosses, epistasis leads to modified F2 phenotypic ratios, such as 9:3:4, 12:3:1, 9:7 or 15:1, instead of the classical 9:3:3:1 ratio.

Mechanistic interpretation

Epistasis often reflects interactions between genes in biochemical pathways or developmental processes:

- One gene controls an earlier step required for the downstream gene to have any effect.
- One gene produces an inhibitor or regulator that blocks the action of another gene.

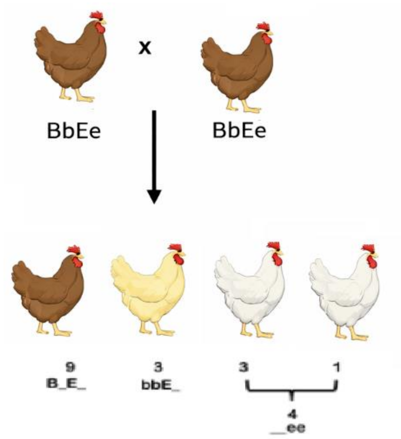
Thus, the epistatic gene determines whether the hypostatic gene’s effect can be seen in the phenotype.

Classical epistatic patterns

- Recessive epistasis (9:3:4): a homozygous recessive genotype at one locus masks the expression of another locus.
- Dominant epistasis (12:3:1): a dominant allele at one locus masks expression of another locus.
- Duplicate recessive epistasis / complementary genes (9:7): at least one dominant allele at both loci is required for the trait to appear.
- Duplicate dominant epistasis (15:1): a dominant allele at either locus produces the same phenotype; only the double recessive differs.

Animal illustration (inhibitor of pigmentation)

In poultry and other species, genes controlling pigment deposition in feathers or hair can be suppressed by inhibitor genes at other loci.
A dominant inhibitor allele may block pigment deposition, so birds carrying this allele are white, even if they possess alleles at other loci that would otherwise produce colored plumage, illustrating epistatic masking of pigment genes.



II.2.3. Sex-linked genes and sex-linked inheritance

II.2.3.1. Definition and general principles

Sex-linked genes are genes located on the sex chromosomes rather than on the autosomes. In mammals, the sex chromosomes are X and Y; in birds, including poultry, they are usually denoted Z and W (or Z and 0 when the W chromosome is absent).
When a gene is present on one sex chromosome and absent from the other, males and females do not carry the same number of copies of that gene, so the inheritance pattern differs between sexes and is described as sex-linked inheritance.

In mammals: females are homogametic (XX) and males are heterogametic (XY), whereas in birds: males are homogametic (ZZ) and females are heterogametic (ZW or Z0).
In poultry, many genes controlling plumage colour, feathering rate, and body size are located on the Z chromosome, so males (ZZ) have two copies of these genes and females (Z0/ZW) have only one copy.

Because of this chromosomal arrangement:

- Recessive Z-linked alleles are more easily expressed in females (who carry only one Z) than in males (who may be heterozygous).
- Crosses often show different phenotypic ratios in sons and daughters, which is a hallmark of sex-linked inheritance in poultry.

II.2.3.2. Importance of sex-linked genes in breeding

Sex-linked genes are particularly valuable in poultry breeding for two main reasons:

1. Autosexing (automatic sex identification)
   Certain sex-linked loci control easily observable traits such as down colour at hatching, feather growth rate or specific plumage patterns.
   By designing appropriate mating schemes, breeders can obtain chicks whose sex is immediately visible at one day of age, which reduces labour, stress and costs associated with manual sexing.

2. Sex-specific control of body size and growth
   Some sex-linked alleles affect mature body size or growth rate.
   Using these alleles allows breeders to reduce the size of parent stock (to save feed and housing space) while keeping normal growth in broilers or layers, thus improving feeding efficiency and overall economic performance.

II.2.3.3. Autosexing genes in poultry

Several Z-linked genes in chickens are used for autosexing because they produce clearly different phenotypes in male and female chicks when specific crosses are made.

Important examples include:

- Rate of feathering (K / k locus)
  One allele leads to slow feathering and the other to rapid feathering.
  By mating slow-feathering males with fast-feathering females (or the reverse), male and female chicks can be distinguished at hatching based on the length of primary wing feathers.

- Barring of plumage (B / b locus)
  A Z-linked allele can produce barred black plumage (alternating light and dark bands), while the recessive allele produces solid black feathers.
  When combined appropriately in parents, male and female chicks show different down patterns, allowing visual sexing in day-old birds.

- Silver vs gold plumage (S / s locus)
  The S allele leads to silver (white) adult plumage and yellow down; the s allele leads to golden plumage and reddish-brown down.
  In certain crosses (for example, silver males × golden females), all male chicks may have yellow down while all female chicks have reddish down, making sexing at one day old very easy and reliable.

By combining these Z-linked alleles in a planned way, commercial hatcheries routinely produce chicks whose sex can be identified at hatching simply by observing down colour or feathering, without invasive manipulation.

II.2.3.4. Sex-linked dwarfism in chickens

A well-known example of a sex-linked gene affecting body size is the dwarfism allele (dw) located on the Z chromosome in chickens.
The recessive allele dw reduces adult body size by about 30–40% compared with normal birds, while the dominant allele DW confers normal size.

Main consequences of using dw in breeding programs:

- Reduced body weight and feed requirements in dwarf females, which lowers feeding costs for parent stock.
- Possibility to house more birds per unit area, since smaller birds require less space, improving housing efficiency.

In practice, the dwarf gene is used in parental lines of broiler production, so that breeding hens are smaller and consume less feed, while the commercial broilers, resulting from specific crosses, still grow to normal market weights.

II.2.4. Lethal and undesirable alleles

II.2.4.1. Lethal alleles: definition and properties

A lethal allele is a mutant form of a gene that causes death when present in a particular genotypic state, most commonly in the homozygous recessive condition (aa), but in some cases in heterozygous dominant form (A_).
Lethal alleles may cause death at different stages: during embryonic development, shortly after birth, or later in life, but in all cases they prevent the affected individuals from reproducing normally.

From a population perspective:

- Recessive lethal alleles can persist for long periods because they are hidden in heterozygous carriers who show no visible defect.
- Dominant lethal alleles are usually eliminated quickly if they cause early death before reproduction; they can only persist if they act late in life or if new mutations arise frequently.

II.2.4.2. Undesirable alleles and genetic defects

An undesirable allele is a gene variant associated with a genetic defect or abnormality that compromises animal health, welfare or economic performance.
Many undesirable alleles are recessive, so carriers appear normal but can produce affected offspring when mated with other carriers; this is especially problematic in populations where a few sires are used extensively.

consequences include:

- Reduced survival or growth of affected animals.
- Increased veterinary costs and management difficulties.
- Loss of embryos or newborns, decreasing reproductive efficiency and farm profitability.

Examples of lethal and undesirable alleles in cattle

1. Example 2

Achondroplasia is a severe skeletal defect in cattle caused by a recessive autosomal allele. Homozygous calves show markedly shortened limbs, deformed skull and facial bones, and usually die before or shortly after birth, often being identified as stillborn or non-viable newborns.

This defect has been reported in the progeny of some widely used artificial insemination sires, illustrating how a single carrier bull can disseminate a harmful allele through many herds.

2. Example 2:

Additional recessive alleles identified in Holstein populations include those responsible for:

- Brachyspina: a shortened and malformed vertebral column often associated with growth retardation and prenatal death.
- Complex vertebral malformation (CVM): a syndrome involving multiple vertebral and limb malformations, leading to embryonic loss or birth of non-viable calves.

The detection of these defects has led to the implementation of routine carrier screening in many breeding organisations, in order to reduce the frequency of these alleles while maintaining genetic progress in production traits.

II.2.4.3. Genetic anomalies sometimes exploited in breeding

Some genetic anomalies, although associated with adverse effects, can also bring economic advantages and are therefore handled cautiously rather than eliminated completely.

A typical example is the double-muscled phenotype in beef cattle, caused by the mh allele:

- Advantages
    - Greatly increased muscle mass and improved carcass conformation, with a higher proportion of high-value cuts.
    - Reduced connective tissue content in muscles, often leading to more tender meat.
- Disadvantages
    - Reduced fertility and maternal abilities in cows carrying the double-muscled genotype.
    - Increased frequency of calving difficulties (dystocia) because of the heavy muscling of calves.
    - Possible reduction in organ size and overall robustness, which may impact animal health and adaptation.

Because of this balance between benefits and risks, breeders may choose to limit the use of the double-muscled allele to specific production systems, or to manage its frequency carefully in order to exploit carcass advantages while avoiding excessive welfare and reproductive problems.

4. "Breed trademarks" and major genes

4.1. Concept of "breed trademarks"

In animal production, certain visible qualitative traits are so characteristic of a breed that they become its "trademark" or signature.
These traits usually have a simple genetic determinism, are easy to recognize on farm, and play a major role in breed identity (for example, horn status, specific coat colour, type of fleece, comb shape in poultry).

Such "trademark" traits often correspond to the action of one or a few major genes with large phenotypic effects, which makes them powerful tools for practical selection and breed differentiation.

4.2. Definition of a major gene

A major gene is a gene whose segregation explains a large part of the variation of a trait in the population, usually with a clear and easily identifiable effect at the individual level.
For qualitative traits, a major gene is often responsible for the presence or absence of a phenotype (horns, a particular colour, a simple defect), whereas for quantitative traits a major gene can create distinct groups of performance within the population.

Major genes differ from the numerous minor genes (polygenes) that each contribute only a small fraction to the variation of quantitative traits such as milk yield or growth rate.

4.3. Characteristics of major genes used as "trademarks"

Major genes that act as breed trademarks generally have the following features:

- Large and easily visible effect: the difference between genotypes is obvious (for example horned vs polled, red vs white, normal vs double-muscled).
- Simple inheritance: usually controlled by one locus with a small number of alleles, following Mendelian segregation (dominant, recessive, codominant or sex-linked).
- High penetrance and stable expression: the phenotype is expressed in most individuals carrying the genotype, with limited environmental influence.
- Clear link to breeding objectives: the trait is directly linked to product differentiation (breed standard, market demand) or to functional objectives (hornlessness for welfare, dwarfism for feed efficiency).

Because of these properties, major genes are key elements in breed descriptions, breeding schemes and communication with farmers and consumers.

4.4. Examples of major genes as breed trademarks

Below are several examples illustrating how major genes define the identity or "brand image" of livestock breeds.

1. Horn status (polled vs horned) in cattle and goats
   The presence or absence of horns is controlled mainly by a major locus, with polled often dominant over horned.
   In some dairy and beef breeds, the widespread use of polled sires has made hornlessness part of the modern breed image and a selling point linked to animal welfare and easier handling.

2. Coat color patterns in beef breeds
   Coat color in many cattle breeds (e.g. black Angus, red Angus, Charolais, Holstein piebald pattern) is largely determined by a few major genes at loci such as Extension, Agouti and spotting loci.
   These color patterns act as instant visual identifiers of the breed and are often specified in breed standards and official labels.

3. Double-muscling in specialized beef breeds
   The muscular hypertrophy ("double-muscled") trait in several beef breeds is due to a major

gene (mh) with a strong effect on muscle development.
In some breeds, a high frequency of the mh allele has become a distinctive trademark associated with very muscular carcasses and high meat yield, even though the gene also has pleiotropic negative effects on fitness and calving.

4. Milk protein variants in dairy breeds
   Major genes coding for milk proteins (for example, kappa-casein, beta-lactoglobulin) have alleles associated with better cheese-making properties.
   Some dairy breeds or lines are promoted as "rich in favorable milk protein variants", making these alleles part of their genetic trademark for the cheese industry.

5. Sex-linked dwarfism and autosexing genes in poultry
   In poultry breeding, the dwarfism gene (dw) and sex-linked color or feathering genes have become trademarks of specific commercial lines used as parent stock, because they contribute to reduced feed consumption and easy sexing of chicks.

4.5. Use of major genes and markers in selection

The identification and characterization of major genes have important practical consequences in modern animal breeding:

- They allow marker-assisted selection, where DNA tests are used to identify carriers of favourable or unfavourable alleles at major loci (polled gene, double-muscling, milk proteins, genetic defects).

- They facilitate introgression of specific traits into a breed by planned crossbreeding, followed by selection for the major gene and backcrossing to the original breed.

- They can be combined with genomic selection on polygenic traits to build composite breeding goals that integrate both "trademark" qualitative traits and quantitative performance.

However, reliance on a small number of major genes must be managed carefully to avoid reducing genetic diversity and to control negative correlated effects (pleiotropy or linkage with undesirable alleles).