Basic Genetics (SQBS 2753)

Extensions of Mendelism

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Beyond Mendel…

• Since Mendel’s work was rediscovered in the early 1900’s:
  – Researchers have studied the many ways genes influence an individual’s phenotype
  – These investigations are called neo-Mendelian genetics (neo from Greek for “new”)
  – This chapter examines types of inheritance observed by researchers that did not conform to the expected Mendelian ratios
Extensions of Mendelian Genetics

- **How alleles affect phenotype**
  - Not always simple dominant/recessive issue
- **Gene interaction**
  - Phenotype controlled by more than one gene
- **Sex-linked genes** (X-linkage in X/Y organisms)
- **Phenotype** can depend on more than genotype
  - Environmental effects
Extended Mendelian Inheritance Patterns

• Incomplete dominance
  – Heterozygosity at a locus produces a third phenotype intermediate to the two homozygous phenotypes

• Co-dominance
  – Heterozygosity at a locus produces a single unique phenotype different from either homozygous condition

• Overdominance
  – Heterozygosity at a locus creates a phenotype that is more beneficial or more detrimental than homozygosity of either locus with any allele
Extended Mendelian Inheritance Patterns

- **Lethality**
  - Homozygosity of an allele kills the cell or organism

- **Penetrance**
  - A measure of how variation in expression of a given allele occurs
  - Incomplete penetrance describes the lack of effect a deleterious allele might have in an individual carrying it
Extended Mendelian Inheritance Patterns

• **Sex-linked**
  – inheritance of genes on that are unique to a sex chromosomes
  – pseudoautosomal genes – genes on both sex chromosomes appear to be on autosomes

• **Sex-influenced**
  – An allele is expressed differently in each sex. Behaving dominantly in one sex and recessively in the other

• **Sex-limited**
  – An allele is only expressed in one or the other sex
EXTENSIONS TO MENDEL FOR SINGLE-GENE INHERITANCE
Complete Dominance/Recessiveness

- recessive allele does *not affect* the phenotype of the heterozygote
- two possible explanations
  - 50% of the normal protein is enough to accomplish the protein’s cellular function
  - The normal gene is “up-regulated” to compensate for the lack of function of the defective allele
Simple Mendelian Inheritance

Normal/dominant allele: P (purple)
Recessive/defective allele: p (white)

<table>
<thead>
<tr>
<th>Genotype</th>
<th>PP</th>
<th>Pp</th>
<th>pp</th>
</tr>
</thead>
<tbody>
<tr>
<td>Amount of functional protein</td>
<td>100%</td>
<td>50%</td>
<td>0%</td>
</tr>
<tr>
<td>Phenotype</td>
<td>Purple</td>
<td>Purple</td>
<td>White</td>
</tr>
</tbody>
</table>
Incomplete Dominance

- **Heterozygote** exhibits a phenotype **intermediate** to the homozygote
- Also called *intermediate dominance* or dosage effect
- Example: Flower colour of snapdragon
- Phenotypic ratio: 1 (red) :2 (pink):1 (white) and **NOT** the 3:1 ratio

<table>
<thead>
<tr>
<th>Phenotype</th>
<th>Genotype</th>
<th>Amount of gene product</th>
</tr>
</thead>
<tbody>
<tr>
<td>Red</td>
<td>RR</td>
<td>2X</td>
</tr>
<tr>
<td>Pink</td>
<td>Rr</td>
<td>X</td>
</tr>
<tr>
<td>White</td>
<td>rr</td>
<td>0</td>
</tr>
</tbody>
</table>
Gene Dosage – A form of intermediate dominance

• Alleles of white –
  – X-linked eye color gene in *Drosophila*
  – W – red (wildtype gene)
  – w - white
  – we - eosin

• we allele was expressed with different intensity in the two sexes
  – Homozygous females ➔ eosin
  – Males ➔ light-eosin
Gene Dosage

• **Morgan & Bridges** hypothesized that difference in intensity was due to the difference in **number of X chromosomes**
  – Female has **two** copies of the “eosin color producer” allele
    • Eyes will contain more color
  – Males have only **one** copy of the allele
    • Eyes will be paler

• This is an example of **gene dosage effect**
Codominance

- two alleles at a locus produce different and detectable gene products in heterozygote
- No dominance or recessiveness
- No “blended” phenotype (not incomplete dominance)

• Example: MN blood group in humans
  - Red blood cell **glycoprotein surface antigen** has two forms (M and N)
  - An individual may exhibit either or both
Codominance

For example:

• One serum (anti-M) recognises only the M antigen; anti-N recognises only N antigen

• Antigen M reacts with anti-M causes AGGLUTINATION

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Phenotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>(L^ML^M)</td>
<td>MM</td>
</tr>
<tr>
<td>(L^ML^N)</td>
<td>MN</td>
</tr>
<tr>
<td>(L^NL^N)</td>
<td>NN</td>
</tr>
</tbody>
</table>
Multiple Alleles

• The term multiple alleles is used to describe situations when three or more different alleles of a gene exist

• Examples:
  – ABO blood
  – Coat color in many species
  – Eye color in Drosophila
Multiple Alleles

- **ABO blood** phenotype is determined by multiple alleles
- ABO type result of antigen on surface of RBCs
  - Antigen A, which is controlled by allele $I^A$
  - Antigen B, which is controlled by allele $I^B$
  - Antigen O, which is controlled by allele $i$

<table>
<thead>
<tr>
<th>Blood Type</th>
<th>O</th>
<th>A</th>
<th>B</th>
<th>AB</th>
</tr>
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<tbody>
<tr>
<td>Genotype</td>
<td>ii</td>
<td>$I^A$I^A or $I^A$i</td>
<td>$I^B$I^B or $I^B$i</td>
<td>$I^A$I^B</td>
</tr>
<tr>
<td>Surface</td>
<td>O</td>
<td>A</td>
<td>B</td>
<td>A and B</td>
</tr>
<tr>
<td>Antigen</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
Allelic Series

• Dominance hierarchy will exist for multiple alleles
  – allelic series for ABO type
    • $I^A = I^B > i$
  – allelic series for rabbit coat color alleles:
    • $C > c^{ch} > c^h > c$
Allelic Series

• **coat color** in rabbits
  - C (full coat color)
  - $c^{ch}$ (chinchilla pattern of coat color)
    • Partial defect in pigmentation
  - $c^h$ (himalayan pattern of coat color)
    • Pigmentation in only certain parts of the body
  - c (albino)
    • Lack of pigmentation
Allelic Series

- Four alleles, c gene in rabbits ---> six heterozygotes;
- $C^+$: completely dominant
- $C^{ch}$ (chinchila allele): partly dominant over the himalayan and albino alleles
- Dominance relationship:

$$C^+ > C^{ch} > C^h > C$$
• *C* gene – formation of black pigment in fur;
• *Albino* allele – nonfunctional allele = null = *amorphic* (completely recessive)
• Partly functional allele = *hypomorphi*c
# Coat Colour in Rabbit

<table>
<thead>
<tr>
<th>Rabbit</th>
<th>Genotype</th>
<th>Phenotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>Albino</td>
<td>CC</td>
<td>White hairs over the entire body</td>
</tr>
<tr>
<td>Himalayan</td>
<td>$C^hC^h$</td>
<td>Black hairs on the extremities; white hairs everywhere else</td>
</tr>
<tr>
<td>Chinchilla</td>
<td>$C^{ch}C^{ch}$</td>
<td>White hair with black tips on the body</td>
</tr>
<tr>
<td>Wild-type</td>
<td>$C^+C^+$</td>
<td>Coloured hairs over the entire body</td>
</tr>
</tbody>
</table>
Lethal Alleles

- **Essential genes** are those that are absolutely required for survival
  - The absence of their protein product leads to a lethal phenotype
    - It is estimated that about 1/3 of all genes are essential for survival

- **Nonessential genes** are those not absolutely required for survival

- A **lethal allele** is one that has the potential to cause the death of an organism
  - These alleles are typically the result of mutations in essential genes
    - usually recessive, but can be dominant
Lethal Alleles

• Example: agouti (coat color) in mice
  – agouti x agouti $\rightarrow$ all agouti
  – yellow x yellow $\rightarrow$ 2/3 yellow, 1/3 agouti
  – agouti x yellow $\rightarrow$ ½ yellow, ½ agouti

  – **Explanation:** mutant yellow dominant over wt agouti and homozygous agouti lethal. Mutant allele always on (gain of function), deletion actually affects neighboring essential gene
Yellow $A^y A^+$ × Yellow $A^y A^+$

\[ A^+ \quad A^y \]
\[ A^+ A^+ \quad (Gray-Brown or agouti) \]
\[ A^y A^y \quad (Yellow) \]
\[ A^+ A^y \quad (Yellow) \]
\[ A^y A^y \quad (Embryonic lethality) \]
Lethal Dominant Mutations

- Both homozygous and heterozygous states are lethal
- Generally very rare
- Example: Huntington disease (humans)
  - Nervous and motor system degeneration
  - Commonly begins to be exhibited after age forty (but can be much earlier)
    - Children already born
- Afflicted persons are heterozygous (Hh)
Conditional Mutations

• The ch allele is a temperature-sensitive conditional mutant
  – The enzyme is only functional at low temperatures
  – Therefore, dark fur will only occur in cooler areas of the body
Overdominance

• **Overdominance** is the phenomenon in which a heterozygote is more vigorous than both of the corresponding homozygotes

• Example:
  – *Sickle-cell heterozygotes* are resistant to malaria
  – increased disease resistance in plant hybrids
Incomplete Penetration

- In some instances, a dominant allele is not expressed in a heterozygote individual
- Example = Polydactyly
  - Autosomal dominant trait
  - Affected individuals have additional fingers and/or toes
  - A single copy of the polydactyly allele is usually sufficient to cause this condition
  - In some cases, however, individuals carry the dominant allele but do not exhibit the trait
Inherited the polydactyly allele from his mother and passed it on to a daughter and son; Does not exhibit the trait himself even though he is a heterozygote.
Incomplete Penetrance

• The term indicates that a dominant allele does not always “penetrate” into the phenotype of the individual

• The measure of penetrance is described at the population level
  – If 60% of heterozygotes carrying a dominant allele exhibit the trait allele, the trait is 60% penetrant

• Note:
  – In any particular individual, the trait is either penetrant or not
Expressivity

• Expressivity is the degree to which a trait is expressed

• In the case of polydactyly, the number of extra digits can vary
  – A person with several extra digits has high expressivity of this trait
  – A person with a single extra digit has low expressivity
Expressivity

• “Eyeless” mutation in *Drosophila*
  – Reduces eye size from a partial reduction to complete elimination (average 0.25 to 0.50)
Penetrance & Expressivity

• The molecular explanation of expressivity and incomplete penetrance may not always be understood

• In most cases, the range of phenotypes is thought to be due to influences of the
  – Environment
  and/or
  – Other genes (genetic background)
Environmental Effects

- **Temperature effects**
  - Evening primrose produces red flowers at 23°C and white flowers at 18°C
  - Siamese cats and Himalayan rabbits have darker fur on cooler areas of body (tail, feet, ears)
    - Enzymes lose catalytic function at higher temperature

- **Temperature sensitive mutations**
  - Mutant allele only expressed (phenotype) at [generally] lower temperature
  - ts phage mutants, restrictive and permissive temperatures

- **Heat-shock genes**
Nutritional Effects

• **Nutritional mutations**
  – Prevent synthesis of nutrient molecules
  – Auxotrophs
  – Phenotype expressed or not depending upon the diet

• **Phenylketonuria (PKU)** – recessive disorder of amino acid metabolism
  – Loss of enzyme to metabolize phenylalanine
  – Severe problems unless low Phe diet

• **Galactosemia** (very bad again) and lactose intolerance (unpleasant)...
Environmental Effects on the Expression of Human Genes

- Pattern baldness – sex-influenced
- Both homo- and heterozygotes – bald patches (male);
- Female – homozygotes – bald (thinning of the hair)
- Relate to testosterone
GENE INTERACTIONS
Epistatic Gene Interactions

- **Gene interactions** occur when two or more different genes influence the outcome of a single trait.
- Most **morphological** traits (height, weight, color) are affected by multiple genes.
- **Epistasis** describes situation between various alleles of two genes.
- **Quantitative loci** is a term to describe those loci controlling quantitatively measurable traits.
- **Pleiotropy** describes situations where one gene affects multiple traits.
Epistasis

• Epistasis
  – The effect of one gene pair (locus) masks or modifies the effect of another gene pair

• Examples
  – Recessive alleles at one locus override expression of alleles at another locus. Alleles at 1st locus are said to be epistatic to the masked hypostatic alleles at the 2nd locus
  – Allele(s) at one locus may require specific allele at another locus, these pairs are said to complement each other
Epistatic Gene Interactions

- examine cases involving 2 loci (genes) that each have 2 alleles
- Crosses performed can be illustrated in general by
  - AaBb X AaBb
    - Where A is dominant to a and B is dominant to b
- If these two genes govern two different traits
  - A 9:3:3:1 ratio is predicted among the offspring
    - simple Mendelian dihybrid inheritance pattern
- If these two genes do affect the same trait the 9:3:3:1 ratio may be altered
  - 9:3:4, or 9:7, or 9:6:1, or 8:6:2 or 12:3:1, or 13:3, or 15:1
    - epistatic ratios
A Cross Producing a 9:7 ratio

**White Variety, $CCpp$** × **White Variety, $ccPP$**

$F_1$ **All purple, $CcPp$**

SELF-FERTILIZATION

$F_2$ progeny

<table>
<thead>
<tr>
<th></th>
<th>$CP$</th>
<th>$Cp$</th>
<th>$cP$</th>
<th>$cp$</th>
</tr>
</thead>
<tbody>
<tr>
<td>$CP$</td>
<td>$CCPP$ Purple</td>
<td>$CCPp$ Purple</td>
<td>$CcPP$ Purple</td>
<td>$CcPp$ Purple</td>
</tr>
<tr>
<td>$Cp$</td>
<td>$CCPp$ Purple</td>
<td>$CCpp$ White</td>
<td>$CcPp$ Purple</td>
<td>$CcPp$ White</td>
</tr>
<tr>
<td>$cP$</td>
<td>$CcPP$ Purple</td>
<td>$CcPp$ Purple</td>
<td>$ccPP$ White</td>
<td>$ccPp$ White</td>
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<tr>
<td>$cp$</td>
<td>$CcPp$ Purple</td>
<td>$CcPp$ White</td>
<td>$ccPp$ White</td>
<td>$ccPp$ White</td>
</tr>
</tbody>
</table>

9 $C_P_ : 3 C_pp : 3 ccP_ : 1 ccpp$

purple : white
**Duplicate Recessive Genes (9:7)**

- When identical phenotypes are produced by both homozygous recessive genotypes, the F1 ratio becomes 9:7.
- The genotype *aaB-, A-bb & aabb* produce one phenotype.
- Both dominant alleles, when present together, complement each other & produce a different phenotype.

- For example:

<table>
<thead>
<tr>
<th>Phenotype Description</th>
<th>Genotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>Flower color of sweet peas</td>
<td>aaB-, A-bb &amp; aabb</td>
</tr>
<tr>
<td><em>A- codes for color pigment</em></td>
<td></td>
</tr>
<tr>
<td><em>B codes for color purple</em></td>
<td></td>
</tr>
<tr>
<td><em>b codes for color white</em></td>
<td></td>
</tr>
</tbody>
</table>
### Duplicate Recessive Genes (9:7)

**P:** \( AAbb \) (white) \( \times \) \( aaBB \) (white)

\[ \text{F}_1: \quad AaBb \) (purple) \]

<table>
<thead>
<tr>
<th>F(_2)</th>
<th>AB</th>
<th>Ab</th>
<th>aB</th>
<th>ab</th>
</tr>
</thead>
<tbody>
<tr>
<td>AB</td>
<td>AABB, purple</td>
<td>AAbb, purple</td>
<td>AaBB, purple</td>
<td>AaBb, purple</td>
</tr>
<tr>
<td>Ab</td>
<td>AAbb, purple</td>
<td>AAbb, white</td>
<td>AaBb, purple</td>
<td>Aabb, white</td>
</tr>
<tr>
<td>aB</td>
<td>AaBB, purple</td>
<td>AaBb, purple</td>
<td>aaBB, white</td>
<td>aaBb, white</td>
</tr>
<tr>
<td>ab</td>
<td>AaBb, purple</td>
<td>Aabb, white</td>
<td>aaBb, white</td>
<td>aabb, white</td>
</tr>
</tbody>
</table>
Epistatic Gene Interaction

• Complementary gene action
  – Enzyme C and enzyme P cooperate to make a product, therefore they complement one another
Epistatic Gene Interaction

- Epistasis describes the situation in which a gene masks the phenotypic effects of another gene.
- Epistatic interactions arise because the two genes encode proteins that participate in sequence in a biochemical pathway.
- If either loci is homozygous for a null mutation, none of that enzyme will be made and the pathway is blocked.

**Diagram:**
- Colorless precursor $\rightarrow$ Enzyme C $\rightarrow$ Colorless intermediate $\rightarrow$ Enzyme P $\rightarrow$ Purple pigment
- For genotype cc, Enzyme C is absent.
- For genotype pp, Enzyme P is absent.
- Colorless pigment is produced when both enzymes are present.

**Genotypes:**
- Genotype cc: Colorless
- Genotype pp: Colorless
A Cross Involving a Two-Gene Interaction Can Still Produce a 9:3:3:1 ratio

• Inheritance of comb morphology in chicken
  – First example of gene interaction
  – William Bateson and Reginald Punnett in 1906
  – Four different comb morphologies:
  – Rose, Pea, Walnut & Single
The crosses of Bateson and Punnett

**Rose comb**
- Wyandotte
  - **RRpp**

**Pea comb**
- Brahma
  - **rrPP**

\[ \text{Rose comb Wyandotte} \quad \times \quad \text{Pea comb Brahma} \]

**F\textsubscript{1}**
- All Walnut,
  - **RrPp**

**SELF-FERTILIZATION**

**F\textsubscript{2}**
- **progeny**

<table>
<thead>
<tr>
<th></th>
<th><strong>RP</strong></th>
<th><strong>Rp</strong></th>
<th><strong>rP</strong></th>
<th><strong>rp</strong></th>
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<tr>
<td><strong>RP</strong></td>
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<td>RrPp Walnut</td>
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<tr>
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<td>RrPp Walnut</td>
<td>rrPP Pea</td>
<td>rrPP Pea</td>
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<tr>
<td><strong>rp</strong></td>
<td>RrPp Walnut</td>
<td>Rppp Rose</td>
<td>rrPP Pea</td>
<td>rppp Single</td>
</tr>
</tbody>
</table>
The crosses of Bateson and Punnett

• $F_2$ generation consisted of chickens with four types of combs
  – 9 walnut : 3 rose : 3 pea : 1 single

• Bateson and Punnett reasoned that comb morphology is determined by two different genes
  – $R$ (rose comb) is dominant to $r$
  – $P$ (pea comb) is dominant to $p$
  – $R$ and $P$ are codominant (walnut comb)
  – $rrpp$ produces single comb
Duplicate Dominant Gene (15:1)

The 9:3:3:1 ratio is modified if the dominant alleles of both loci each produce the same phenotype without cumulative effect.

For example:

Flower color of peas

- `aabb` codes for color white
- any other combination produce color red

<table>
<thead>
<tr>
<th>P:</th>
<th>AAbb (red)</th>
<th>X</th>
<th>aaBB (white)</th>
</tr>
</thead>
<tbody>
<tr>
<td>F₁:</td>
<td>AaBb (red)</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>F₂</th>
<th>AB</th>
<th>Ab</th>
<th>aB</th>
<th>ab</th>
</tr>
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<tbody>
<tr>
<td>A</td>
<td>AABB, red</td>
<td>AAbb, red</td>
<td>AaBB, red</td>
<td>AaBb, red</td>
</tr>
<tr>
<td>B</td>
<td>AABB, red</td>
<td>AAbb, red</td>
<td>AaBB, red</td>
<td>AaBb, red</td>
</tr>
<tr>
<td>Ab</td>
<td>AABb, red</td>
<td>AAbb, red</td>
<td>AaBb, red</td>
<td>Aabb, red</td>
</tr>
<tr>
<td>aB</td>
<td>AaBB, red</td>
<td>AaBb, red</td>
<td>aaBB, red</td>
<td>aaBb, red</td>
</tr>
<tr>
<td>ab</td>
<td>AaBb, red</td>
<td>Aabb, red</td>
<td>aaBb, red</td>
<td>aabb, white</td>
</tr>
</tbody>
</table>
Gene Interaction

• Duplicate gene action
  – Enzyme 1 and enzyme 2 are redundant
  – They both make product C, therefore they duplicate each other
Dominant Epistasis (12:3:1)

• When the dominant allele (A) produces a certain phenotype regardless of the allele condition of another locus (B), A is said to be epistatic to B.

♀ The dominant allele A is able to express itself in the presence of either B or b.
♀ Only when the genotype of the individual is homozygous recessive (aa), then B or b can be expressed.
♀ A-B- & A-bb produce the same phenotype;
♀ aaB- & aabb produce 2 additional phenotypes.
Dominant Epistasis (12:3:1)

For example:

Coat colors of dogs

I - inhibit coat color pigment / expression

B represents **black** color coat

b represents **brown** color coat

P:  \( liBb \) (white)  \( \times \)  \( liBb \) (white)

F\(_1\):  \( liBb \)

<table>
<thead>
<tr>
<th>( F_2 )</th>
<th>( iB )</th>
<th>( ib )</th>
<th>( iB )</th>
<th>( ib )</th>
</tr>
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<tbody>
<tr>
<td>( iB )</td>
<td>( liBB, ) white</td>
<td>( liBb, ) white</td>
<td>( liBB, ) white</td>
<td>( liBb, ) white</td>
</tr>
<tr>
<td>( Ib )</td>
<td>( liBb, ) white</td>
<td>( libb, ) white</td>
<td>( liBb, ) white</td>
<td>( libb, ) white</td>
</tr>
<tr>
<td>( iB )</td>
<td>( liBB, ) white</td>
<td>( liBb, ) white</td>
<td>( iiBB, ) black</td>
<td>( iibb, ) black</td>
</tr>
<tr>
<td>( ib )</td>
<td>( liBb, ) white</td>
<td>( libb, ) white</td>
<td>( iiBb, ) black</td>
<td>( iibb, ) brown</td>
</tr>
</tbody>
</table>
Recessive Epistasis (9:3:4)

- If the recessive genotype at locus A (eg: \(aa\)) suppresses the expression of alleles at B locus, **locus A exhibit recessive epistasis over locus B**.

- The alleles in **locus B can only be expressed** with the presence of dominant alleles at locus A.

- Genotypes **A-B- & A-bb** produce 2 additional phenotypes.

- For example:
  - **Flower color of peas**
  - \(A-\) codes for color pigment
  - \(B\) codes for color **purple**
  - \(b\) codes for color **red**
Recessive Epistasis (9:3:4)

P:  \( AAbb \) (red) \( \times \) \( aaBB \) (white)

\( F_1: \quad AaBb \) (purple)

<table>
<thead>
<tr>
<th>F_2</th>
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<th>Ab</th>
<th>aB</th>
<th>ab</th>
</tr>
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<tbody>
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<td>AaBB, purple</td>
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</tr>
<tr>
<td>Ab</td>
<td>AABb, purple</td>
<td>Abb, red</td>
<td>AaBb, purple</td>
<td>Aabb, red</td>
</tr>
<tr>
<td>aB</td>
<td>AaBB, purple</td>
<td>AaBb, purple</td>
<td>aaBB, white</td>
<td>aaBb, white</td>
</tr>
<tr>
<td>ab</td>
<td>AaBb, purple</td>
<td>Abb, red</td>
<td>aaBb, white</td>
<td>aabb, white</td>
</tr>
</tbody>
</table>

**Table Legend:**
- Purple: Allele combination results in purple.
- Red: Allele combination results in red.
- White: Allele combination results in white.
Duplicate Genes with Cumulative Effect

(9:6:1)

- Occur when dominant allele (homozygous or heterozygous) at either locus (but not both) produces the same phenotype.
- Genotypes $A-bb$ & $aaB-$ produce one unit each and therefore have the same phenotype.
- Genotype $aabb$ produces no pigment but in genotype $A-B-$ the effect is cumulative and 2 units of phenotypes are produced.

- For example:
  - **Color of wheat kernels**
    - $R-B-$ produce red color
    - $rrbb$ produce white color
    - Any other combination produces brown color

.
### P: RRBB (red) X rrbb (white)

### F₁: RrBb (red)

### F₂:

<table>
<thead>
<tr>
<th></th>
<th>RB</th>
<th>Rb</th>
<th>rB</th>
<th>rb</th>
</tr>
</thead>
<tbody>
<tr>
<td>RB</td>
<td>RRBB, red</td>
<td>RRBb, red</td>
<td>RrBB, red</td>
<td>RrBb, red</td>
</tr>
<tr>
<td>Rb</td>
<td>RRBb, red</td>
<td>RRbb, brown</td>
<td>RrBb, red</td>
<td>Rrbb, brown</td>
</tr>
<tr>
<td>rB</td>
<td>RrBB, red</td>
<td>RrBb, red</td>
<td>rrBB, brown</td>
<td>rrBb, brown</td>
</tr>
<tr>
<td>rb</td>
<td>RrBb, red</td>
<td>Rrbb, brown</td>
<td>rrBb, brown</td>
<td>rrbb, white</td>
</tr>
</tbody>
</table>
Dominant and Recessive Interaction (13:3)

✓ Only two F2 phenotypes result when a dominant genotype at 1 locus (A-) and the recessive genotype at another (bb) produce the same phenotypic effect.
✓ Genotype A-B-, aaB- & aabb produce one phenotype and genotype A-bb produce another in the ratio 13:3.

<table>
<thead>
<tr>
<th>F2</th>
<th>AB</th>
<th>Ab</th>
<th>aB</th>
<th>ab</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>ABB, white</td>
<td>AABb, white</td>
<td>AaBB, white</td>
<td>AaBb, white</td>
</tr>
<tr>
<td>AB</td>
<td>AABb, white</td>
<td>AAbb, red</td>
<td>AaBb, white</td>
<td>Aabb, red</td>
</tr>
<tr>
<td>Ab</td>
<td>AABb, white</td>
<td>AAbb, red</td>
<td>AaBb, white</td>
<td></td>
</tr>
<tr>
<td>aB</td>
<td>AaBB, white</td>
<td>AaBb, white</td>
<td>aaBB, white</td>
<td>aaBb, white</td>
</tr>
<tr>
<td>ab</td>
<td>AaBb, white</td>
<td>Aabb, red</td>
<td>AaBb, white</td>
<td>aabb, white</td>
</tr>
</tbody>
</table>
Dominant and Recessive Interaction (13:3)

❄ Only two F2 phenotypes result when a dominant genotype at 1 locus (A-) and the recessive genotype at another (bb) produce the same phenotypic effect.
❄ Genotype A-B-, aaB- & aabb produce one phenotype and genotype A-bb produce another in the ratio 13:3.
Dominant and Recessive Interaction (13:3)

For example:

Flower color of peas

A- bb codes for color red

Any other combination codes for color white

P: $AAbb$ (white) $\times$ $aaBB$ (white)
F$_1$: $AaBb$ (white)

<table>
<thead>
<tr>
<th></th>
<th>$AB$</th>
<th>$Ab$</th>
<th>$aB$</th>
<th>$ab$</th>
</tr>
</thead>
<tbody>
<tr>
<td>$AB$</td>
<td>$AABB$, white</td>
<td>$AABb$, white</td>
<td>$AaBB$, white</td>
<td>$AaBb$, white</td>
</tr>
<tr>
<td>$Ab$</td>
<td>$AABb$, white</td>
<td>$AAbb$, red</td>
<td>$AaBb$, white</td>
<td>$Aabb$, red</td>
</tr>
<tr>
<td>$aB$</td>
<td>$AaBB$, white</td>
<td>$AaBb$, white</td>
<td>$aaBB$, white</td>
<td>$aaBb$, white</td>
</tr>
<tr>
<td>$ab$</td>
<td>$AaBb$, white</td>
<td>$Aabb$, red</td>
<td>$aaBb$, white</td>
<td>$aabb$, white</td>
</tr>
</tbody>
</table>
## Summary of Epistatic Ratios

<table>
<thead>
<tr>
<th>Genotypes</th>
<th>A-B-</th>
<th>A-bb</th>
<th>aaB-</th>
<th>aabb</th>
</tr>
</thead>
<tbody>
<tr>
<td>Classical ratio</td>
<td>9</td>
<td>3</td>
<td>3</td>
<td>1</td>
</tr>
<tr>
<td>Dominant epistasis</td>
<td>12</td>
<td>3</td>
<td></td>
<td>1</td>
</tr>
<tr>
<td>Recessive epistasis</td>
<td>9</td>
<td>3</td>
<td>4</td>
<td></td>
</tr>
<tr>
<td>Duplicate genes with cumulative effect</td>
<td>9</td>
<td>6</td>
<td></td>
<td>1</td>
</tr>
<tr>
<td>Duplicate dominant genes</td>
<td></td>
<td>15</td>
<td></td>
<td>1</td>
</tr>
<tr>
<td>Duplicate recessive genes</td>
<td>9</td>
<td></td>
<td>7</td>
<td></td>
</tr>
<tr>
<td>Dominant and recessive interaction</td>
<td>13</td>
<td>3</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
## Examples of Epistatic Cases

<table>
<thead>
<tr>
<th>Organism</th>
<th>Character</th>
<th>F2 Phenotypes</th>
<th>Modified ratio</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mouse</td>
<td>Coat colour</td>
<td>Agouti</td>
<td>albino</td>
</tr>
<tr>
<td>Squash</td>
<td>Colour</td>
<td>White</td>
<td>Yellow</td>
</tr>
<tr>
<td>Pea</td>
<td>Flower colour</td>
<td>Purple</td>
<td>White</td>
</tr>
<tr>
<td>Squash</td>
<td>Fruit shape</td>
<td>Disc</td>
<td>Sphere</td>
</tr>
<tr>
<td>Chicken</td>
<td>Colour</td>
<td>White</td>
<td>Coloured</td>
</tr>
</tbody>
</table>
References

