

Complex Patterns of Inheritance



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التواصل الاجتماعي بحسب الصف الحادي عشر العام



صفحة المناهج
الإماراتية على
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المزيد من الملفات بحسب الصف الحادي عشر العام والمادة علوم في الفصل الثالث

Basic Patterns of Human Inheritance: Pedigrees

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Applied Genetics

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Genetic Recombination and Gene Linkage

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Mendelian Genetics

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Incomplete Dominance

When an organism is heterozygous for a trait, its phenotype is usually that of the dominant trait.

Example: A pea plant with genotype Tt (T = tall allele) will be tall.

Example: Snapdragons

- Red-flowered snapdragons: Genotype = $C^R C^R$
- White-flowered snapdragons: Genotype = $C^W C^W$
- Crossing them : Heterozygous offspring: Genotype = $C^R C^W$
 - These offspring have pink flowers → **Figure 18**

Definition

Incomplete dominance is when the heterozygous phenotype is intermediate between the two homozygous phenotypes.

F₁ Generation

- If $C^R C^W$ (pink) plants are self-fertilized:
 - Offspring flower colors appear in a 1:2:1 ratio:
 - Red : Pink : White

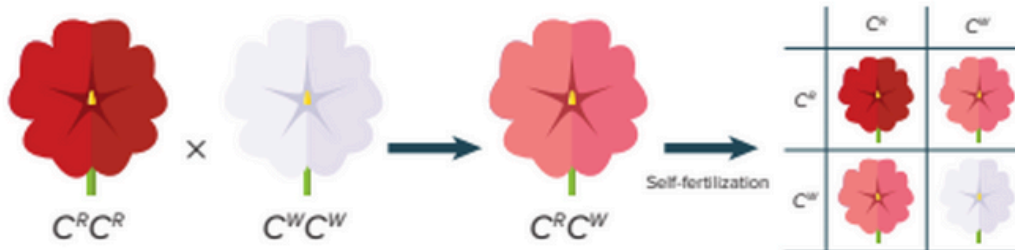


Figure 18

- The color of snapdragon flowers is an example of incomplete dominance.
- When:
 - A white-flowered plant is crossed with a red-flowered plant,
 → The offspring have pink flowers.
- When pink-flowered plants self-fertilize, the offspring include:
 - Red, Pink, White
 - In a 1:2:1 ratio.

Predict:

What would happen if you crossed a pink-flowered snapdragon with a white-flowered snapdragon? Identify the genotypes and phenotypes possible in the offspring.

- Genotypes: 50% $C^R C^W$ (pink), 50% $C^W C^W$ (white)
- Phenotypes: 50% Pink, 50% White



Codominance

Normally, when an organism is heterozygous for a trait, the dominant phenotype is expressed. However, in a complex inheritance pattern called **codominance**: Both alleles are expressed in the heterozygous condition.

Example:

Sickle-cell disease in humans is a case study of codominant inheritance.

Sickle-cell Disease

Prevalence

- Particularly common in people of African descent.
- About 9% of African Americans carry one form of the trait.

What It Affects

- Affects red blood cells and their ability to transport oxygen.

Visual Reference

- **Figure 19:**
 - **Middle image:** shows blood cells of a heterozygous individual.
 - **Top image:** shows a normal red blood cell.

Cause

- Caused by changes in hemoglobin (protein in red blood cells).
- This change causes cells to become sickle-shaped (C-shaped).

Impact

1-Sickle-shaped cells:

- Do not transport oxygen efficiently.
- Block circulation in small vessels.

2-Heterozygous individuals:

- Have both normal and sickle-shaped cells.
- Can lead relatively normal lives, as normal cells compensate.



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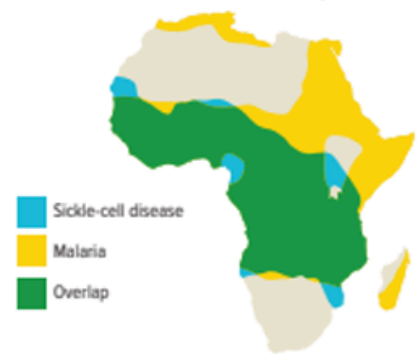


Figure 19

- **Top:**

Normal red blood cells are flat and disk-shaped.

- **Middle:**

Sickle-shaped cells are elongated and/or C-shaped. They can clump, blocking circulation in small vessels.

- **Bottom:**

The sickle-cell allele increases resistance to malaria.



Sickle-cell Disease and Malaria

- In **Figure 19**, the bottom image shows:
 - The distribution of sickle-cell disease and malaria in Africa.
 - Many areas with sickle-cell disease overlap with malaria regions.

Why is the sickle-cell allele common in central Africa?

- People who are heterozygous for the sickle-cell trait have:
 - Higher resistance to malaria

Scientific Insight:

- In areas with high malaria:
 - The death rate due to malaria is lower among people carrying the sickle-cell allele.
 - This allows more survivors, who pass on the allele to the next generation.

Result:

- Sickle-cell disease continues to increase in Africa, particularly where malaria is widespread.



Multiple Alleles

- Not all traits are determined by just two alleles.
- Some traits are controlled by **more than two alleles** — called multiple alleles.

Example: Human blood group

Blood Groups in Humans

The ABO blood group (shown in **Figure 20**) has three alleles:

- I^A → Blood type A
- I^B → Blood type B
- i → Blood type O (absence of A/B markers)

Genotypes & Blood Types:

- ii → Type O
- $I^A I^A$ or $I^A i$ → Type A
- $I^B I^B$ or $I^B i$ → Type B
- $I^A I^B$ → Type AB

Allele Relationships

- i is recessive to both I^A and I^B
- I^A and I^B are codominant
 - → That's why AB blood type occurs when both alleles are inherited ($I^A I^B$)



Conclusion:

The ABO blood group is an example of both:

- Multiple alleles
- Codominance



Rh Blood Group System

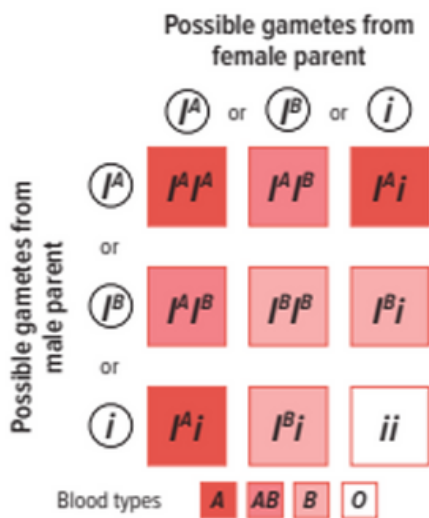
- **Rh factors are inherited from both parents.**
- **They determine whether blood type is:**
 - Rh⁺ (positive)
 - Rh⁻ (negative)

Inheritance Pattern

- Rh⁺ = Dominant
- Rh⁻ = Recessive

Origin of the Name

- The Rh factor is a blood protein.
- It was named after the rhesus monkey, because:
 - The protein was first discovered during studies on rhesus monkeys.



	I^A (♀)	I^B (♀)	i (♀)
I^A (♂)	$I^A I^A$ (A)	$I^A I^B$ (AB)	$I^A i$ (A)
I^B (♂)	$I^A I^B$ (AB)	$I^B I^B$ (B)	$I^B i$ (B)
i (♂)	$I^A i$ (A)	$I^B i$ (B)	ii (O)

Figure 20

Three forms of alleles Involved:

- $I^A \rightarrow$ Blood type A
- $I^B \rightarrow$ Blood type B
- $i \rightarrow$ Blood type O (no markers)

Dominance Rules:

- I^A and I^B are codominant
- i is recessive to both



Coat Color in Rabbits

(Example of multiple alleles and dominance hierarchy)

Alleles Involved:

- $C \rightarrow$ Full color (dominant to all others)
- $c^{ch} \rightarrow$ Chinchilla (dominant to c^h and c)
- $c^h \rightarrow$ Himalayan (dominant to c)
- $c \rightarrow$ Recessive to all alleles. Produces **albino** phenotype only when homozygous (cc).

Dominance hierarchy:

- $C > c^{ch} > c^h > c$

Figure 21:

- Shows the genotypes and phenotypes for rabbit coat color.
- Full color is dominant over chinchilla, which is dominant over Himalayan, which is dominant over albino.

Why This Matters (Genetic Variation Insight)

- Without multiple alleles (e.g. with just T and t), we only get:
 - 3 genotypes: TT , Tt , tt
 - 2 phenotypes (e.g. tall or short)

With 4 rabbit coat alleles, we get:

- 10 different genotypes
- 4 phenotypes

(based on which allele dominates in the pair)

Why More Variation?

- The presence of multiple alleles expands the genotype/phenotype possibilities.
- In rabbits, coat color variation also increases due to interactions of the color gene with other genes.



Full color
 CC or Cc or Cc^h or Cc^{ch}



Chinchilla
 $c^{ch}c^{ch}$ or $c^{ch}c^h$ or $c^{ch}c$



Himalayan
 $c^h c^h$ or $c^h c$



Albino
 cc

Figure 21 Rabbits have multiple alleles for coat color. The four alleles provide four basic variations in coat color.

**N.B:****Rabbit Coat Color – Figure 21**

Each coat color is determined by a combination of 4 alleles:

- C (Full color – most dominant)
- c^{ch} (Chinchilla)
- c^h (Himalayan)
- c (Albino – most recessive)

Phenotype	Genotypes	Notes
Full Color	CC, Cc, Cc^{ch} , Cc^h	Dominant over all others
Chinchilla	$c^{ch}c^{ch}$, $c^{ch}c^h$, $c^{ch}c$	Dominant over Himalayan and Albino
Himalayan	$c^h c^h$, $c^h c$	Dominant over albino only
Albino	cc	Only expressed when homozygous recessive

Epistasis

(Pronounced: ih-PIHS-tuh-sus)

Definition

- **Epistasis** = when one gene masks or hides the effect of another gene.
- In Labradors: the E/e gene determines if pigment is produced, while the B/b gene decides how dark the pigment is.

How it works

- E allele (dominant) → allows pigment (dark pigment).
- ee genotype → no pigment at all → coat is yellow regardless of B/b alleles.
- B allele (dominant) → controls darkness of pigment (B = black, b = brown).

Genotypes and Phenotypes

- eeBB, eeBb, eebb → Yellow (no pigment, E gene blocked).
- E_bb → Chocolate brown.
- E_B_ → Black (at least one E and one B).



Figure 22

- The results of epistasis in coat color in Labrador retrievers
- Show an interaction of two genes,
- Each with two alleles

Dosage compensation

Human females have:

- 22 pairs of autosomes
- 1 pair of X chromosomes

Males have:

- 22 pairs of autosomes
- 1 X and 1 Y chromosome
- The X chromosome is larger than the Y chromosome.
- The X chromosome carries genes needed for development in both females and males.
- The Y chromosome mainly carries genes for development of male characteristics.

Dosage Compensation (X-Inactivation)

- Females have two X chromosomes → seem to get double dose of X-linked genes.
- Males have only one X → get one dose.
- **To balance this:**
 - One of the X chromosomes in females stops working in each body cell.
 - This process is called dosage compensation or X-inactivation.
 - The inactive X is chosen randomly in each cell.
 - This occurs in all mammals.

Human Genome Project Insight

- **The NIH (National Institutes of Health) discovered:**
 - New info about the X chromosome sequence.
 - Some genes on the inactivated X may be more active than previously believed.



Chromosome Inactivation

- The coat colors of the calico cat (shown in **Figure 23**) are caused by:
 - Random inactivation of a particular X chromosome.
- The resulting colors depend on:
 - Which X chromosome is activated.

Color Pattern Explanation:

- **Orange patches:**
 - Formed by inactivation of the X chromosome carrying the black coat color allele.
- **Black patches:**
 - Result from inactivation of the X chromosome carrying the orange coat color allele.

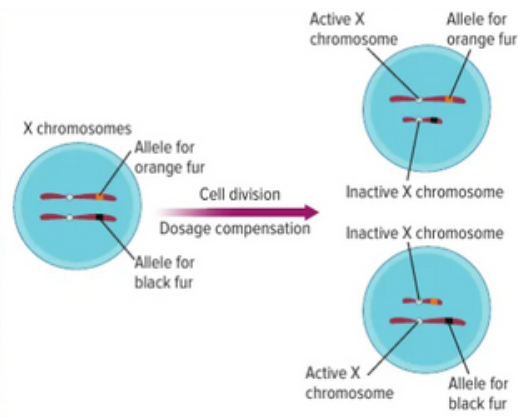


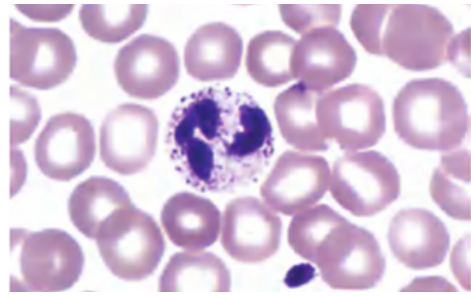
Figure 23

- The calico coat of this cat results from the random inactivation of the X chromosomes in body cells.
- One X chromosome codes for orange fur, and one X chromosome codes for black fur,
- As illustrated on the right.

Barr Bodies

- The inactivated X chromosomes can be observed in cells.
- In 1949, Canadian scientist Murray Barr:
 - Observed inactivated X chromosomes in female calico cats.
 - Noticed a condensed, darkly stained structure in the nucleus.
- The darkly stained, inactivated X chromosomes (like the one shown in Figure 24) are called Barr bodies.
- It was later discovered that:

Only females, including human females, have Barr bodies in their cell nuclei.

**Figure 24**

- An inactivated X chromosome in a female body cell is called a Barr body.
- It is a dark body, usually found near the nucleus.

Sex-Linked Traits

- **Traits controlled by genes located on the X chromosome are called sex-linked traits, or X-linked traits.**
- **Because genetically classified males have only one X chromosome, they:**
 - Are affected by recessive X-linked traits more often than genetically classified females.
- **Females are less likely to express a recessive X-linked trait because:**
 - The other X chromosome may mask the effect of the trait.

Autosomal Traits That Appear Sex-Linked

- **Some traits located on autosomes may appear sex-linked, even though they are not.**
- **This occurs when:**
 - An allele appears dominant in one sex, but recessive in the other.

Example: Male-Pattern Baldness

- **The allele for baldness is:**
 - Recessive in females
 - Dominant in males, causing hair loss in a typical pattern
- A male would be bald if he were heterozygous for the trait.
- A female would be bald only if she were homozygous recessive.



Red-green colorblindness

- The trait for red-green colorblindness, most commonly, is a recessive X-linked trait.
- About 8 percent of males in the United States have this trait.
- **The right photo in Figure 25 shows:**
 - How a person with red-green colorblindness might view colors.
- **Compared to:**
 - The left photo, which shows how colors appear to a person without red-green colorblindness.



Punnett Square – Figure 25

- **The mother is a carrier for color blindness:**
 - She has the recessive allele on one of her X chromosomes.
- **The father is not color blind:**
 - He does not have the recessive allele on his one X chromosome.
- **The sex-linked trait is shown by:**
 - Writing the allele on the X chromosome.

Offspring Outcomes

- **The only offspring that can possibly have red-green color blindness is:**
 - A male child.
- It is also possible for male offspring to have normal vision.
- Because it is an X-linked trait,

Red-green colorblindness is very rare in females.



Normal vision



Red-green color blindness (shades of gray)

X^B = Normal
 X^b = Red-green color blindness
 Y = Y chromosome

Dad

	X^B	Y
X^B	$X^B X^B$	$X^B Y$
X^b	$X^B X^b$	$X^b Y$

Mom

Color blind male

Figure 25

People with red-green color blindness view red and green as shades of gray.

Explain why there are fewer females who have red-green color blindness than males.

Answer:

- Males only need one copy of the X^b allele ($X^b Y$) to be color blind.
- Females must inherit two copies ($X^b X^b$), one from each parent, to be color blind.
- This makes red-green color blindness more common in males and rare in females.



Hemophilia

- Hemophilia is another recessive sex-linked disorder.
- Characterized by delayed clotting of the blood.
- Like red-green color blindness, this disorder is more common in males than females.

Historical Example: Queen Victoria's Family

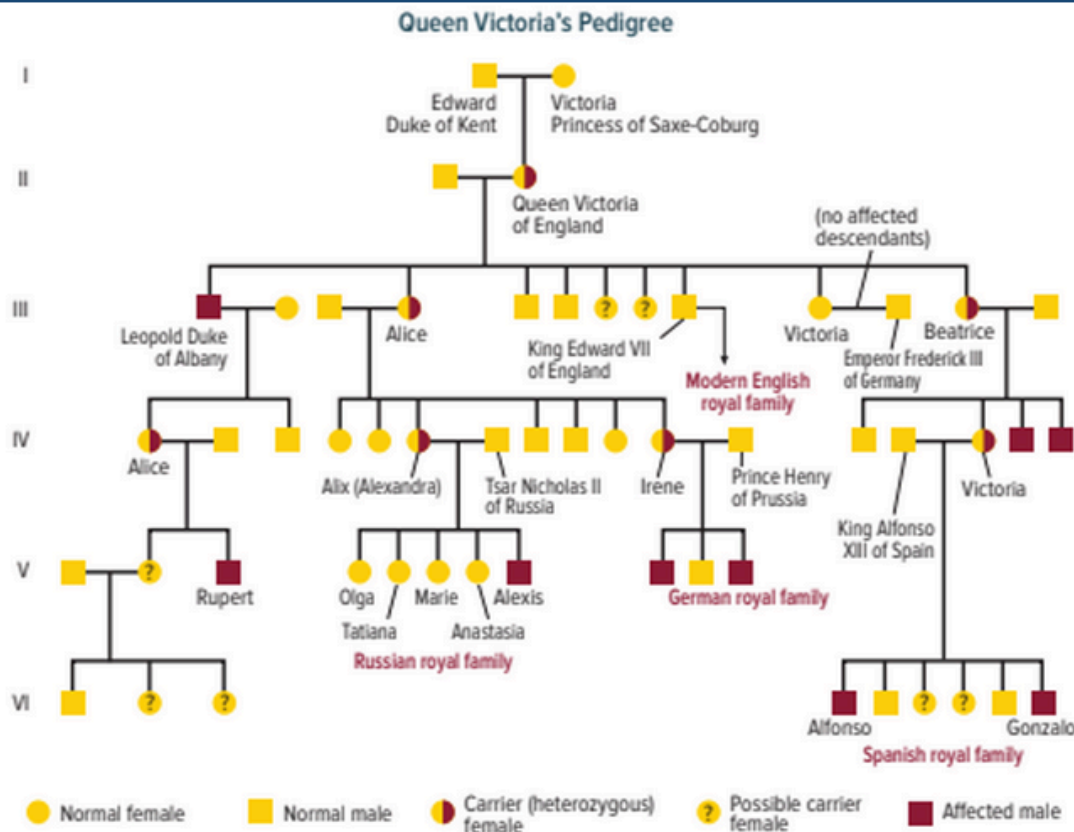
- **A famous pedigree of hemophilia arose in the family of Queen Victoria of England (1819–1901).**
 - Her son Leopold died of hemophilia.
 - Her daughters Alice and Beatrice were carriers for the disease (**as shown in Figure 26**).
 - Alice and Beatrice passed hemophilia to the Russian, German, and Spanish royal families.
- **Generational transmission:**
 - Alexandra (granddaughter, carrier) → married Tsar Nicholas II of Russia.
 - Irene (granddaughter) → passed trait to German royal family.
 - Victoria (granddaughter) → passed trait to Spanish royal family.

Medical Impact

- Men with hemophilia usually died at an early age until the twentieth century.
- Discovery of clotting factors → allowed treatment of hemophiliacs.
- Problem: blood-borne viruses (Hepatitis C, HIV) were often transmitted to hemophiliacs through transfusions until the 1990s.
- Solution: safer methods of blood transfusion were developed in the 1990s.

Summary:

- Hemophilia was inherited through carrier females.
- Disease spread to:
 - Russian Royal Family (Alexis affected)
 - German Royal Family (affected sons)
 - Spanish Royal Family (Alfonso and Gonzalo affected)
- Modern English Royal Family (via Edward VII) had no affected descendants.

**Figure 26**

The pedigree above shows the inheritance of hemophilia in the royal families of England, Germany, Spain, and Russia, starting with the children of Queen Victoria.

**N.B:****II Generation**

- Queen Victoria of England = (Carrier)
- Her parents: Edward Duke of Kent & Victoria Princess of Saxe-Coburg

III Generation (Her Children):

1. Leopold Duke of Albany = (Affected male)
2. Alice = (Carrier)
3. King Edward VII = (Normal male)
4. Victoria = (Normal female)
5. Beatrice = (Carrier)
6. Others = (Unknown carrier status)

Generation IV

- Alice's lineage (linked to Russian Royal Family):
 - Daughter: Alix (Alexandra) – Carrier
 - Married Tsar Nicholas II (Normal male)
 - Their son: Alexis – Affected male
 - Daughters: Normal females
- Irene's lineage (linked to German Royal Family):
 - Irene – Carrier
 - Married Prince Henry of Prussia (Normal male)
 - Sons: Some affected males
- Beatrice's lineage (linked to Spanish Royal Family):
 - Beatrice – Carrier
 - Married King Alfonso XII of Spain (Normal male)
 - Sons: Alfonso and Gonzalo – Both affected
 - Some daughters: Possible carriers



Polygenic Traits

- You have examined traits determined by a pair of genes.
- Many phenotypic traits, however, arise from the interaction of multiple pairs of genes.
- Such traits are called **polygenic traits**.

Examples of polygenic traits:

- Skin color
- Height
- Eye color
- Fingerprint pattern

Characteristic of polygenic traits:

- When the frequency of the number of dominant alleles is graphed (as shown in Figure 27), the result is a bell-shaped curve.
- This shows that:
 - More intermediate phenotypes exist
 - Compared to extreme phenotypes

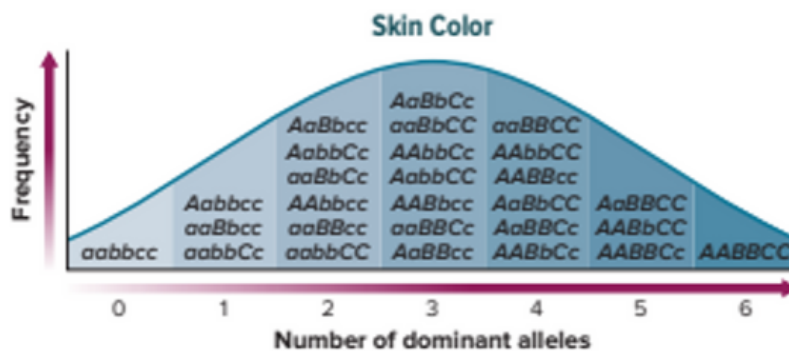


Figure 27

- This graph shows possible shades of skin color from three sets of alleles,
- although the trait is thought to involve more than three sets of alleles.

Predict

Would more gene pairs increase or decrease the number of possible phenotypes?

More gene pairs would increase the number of possible phenotypes.



Environmental Influences

Variation and Distribution of Traits

- **Traits in a population depend on:**

- a. Genetic factors
- b. Environmental factors

Role of Environmental Factors

- Environmental factors affect the expression of traits.
- They influence the probability of traits appearing in a population.

Example: Heart Disease

- Inheritance: Tendency to develop heart disease can be inherited.
- Environmental influence:
 - Diet
 - Exercise
- These factors contribute to the occurrence and seriousness of the disease.

Common Environmental Influences on Phenotype

- Sunlight
- Water
- Temperature



Figure 28

- Temperature affects the expression of color pigment in the fur of Siamese cats.



Sunlight and water

- Without enough sunlight, most flowering plants do not bear flowers.
- Many plants lose their leaves in response to water deficiency.



Temperature

- Most organisms experience phenotypic changes from extreme temperature changes.

In extreme heat, for example:

- Many plants' leaves droop.
- Flower buds shrivel.
- Chlorophyll disappears.
- Roots stop growing.



These examples probably do not surprise you, although you may have never thought of them as phenotypic changes.

Temperature also influences the expression of genes.

Example:

Notice the fur of the Siamese cat shown in **Figure 28**.

- The gene that codes for production of the color pigment in the Siamese cat's body functions only under cooler conditions.
- Therefore:
 - Cooler regions are darker.
 - Warmer regions, where pigment production is inhibited by temperature, are lighter.

Twin Studies

Identical twins are genetically the same.

If a trait is inherited, both twins will have it.

Scientists conclude:

- Traits that appear frequently in both identical twins are at least partially due to heredity.
- Traits expressed differently in identical twins are strongly influenced by environment.
- The percentage of twins who both express a given trait

is called a **concordance rate**.

- **Figure 29** shows some traits and their concordance rates.

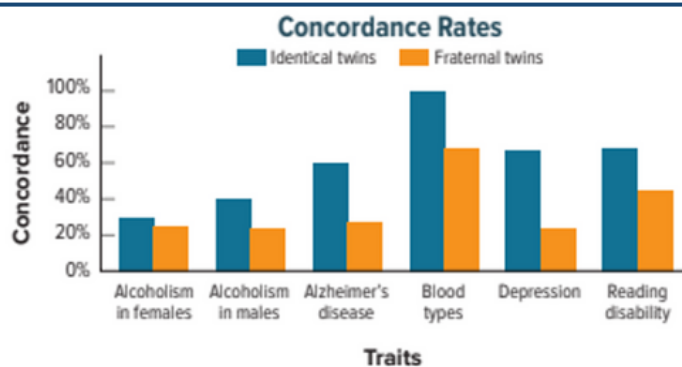


Figure 29:

When a trait is found more often in both members of identical twins than in fraternal twins, the trait is presumed to have a significant inherited component.

- Traits with higher concordance in identical vs fraternal twins → more heritable.
- Traits with low concordance even in identical twins → more environmentally influenced.
- Blood type is mostly genetic.
- Mental/behavioral traits (e.g., depression, alcoholism) are influenced by both genes and environment.