

بِسْمِ اللّٰهِ الرَّحْمٰنِ الرَّحِیْمِ

Single-Gene Inheritance

Comprehensive study sheet based on the uploaded lecture slides

Importance of Family History



- Understanding the past is the key to predicting the future.

Slide 2: family history helps predict future risk.

Built from every slide in the lecture, with the main concepts reorganized into a cleaner study format.

1. Big picture and objectives

- The lecture starts with a simple idea: understanding the past is the key to predicting the future.
- Family history is useful because inherited patterns repeat across generations.

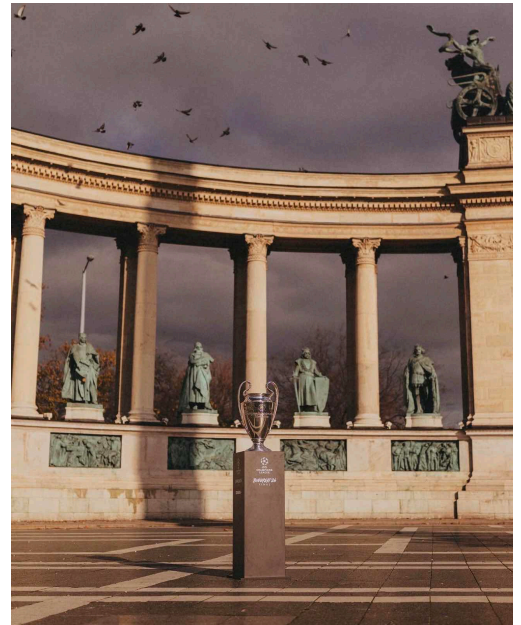
Lecture objectives

- Construct and interpret pedigrees using standard nomenclature.
- Describe the general features of Mendelian patterns of single-gene inheritance.
- Identify the mode of inheritance of traits discussed in lecture.
- Describe aspects of phenotypic expression using the lecture examples.
- Understand basic concepts of probability.
- Recognize the pattern of inheritance of a trait segregating in a family.
- Apply probability and Mendelian principles to calculate affected and unaffected offspring.

Concept 14.3 in one idea

- Inheritance is often more complex than the pea-plant examples Mendel studied.
- Many heritable traits are not controlled by just one gene with two alleles.
- Still, segregation and independent assortment remain the foundation.

- The relationship between genotype and phenotype is rarely as simple as in the pea plant characters Mendel studied
- Many heritable characters are not determined by only one gene with two alleles
- However, the basic principles of segregation and independent assortment apply even to more complex patterns of inheritance



متى تنصفين كبير إسبانيا يا أمجد البطولات

2. Ways single-gene inheritance can differ from the simplest Mendelian model

- A single gene can show non-classic inheritance when alleles are not completely dominant or recessive, when a gene has more than two alleles, or when one gene produces multiple phenotypes.

Degrees of dominance

Pattern	Core idea	Example
Complete dominance	Heterozygote = dominant homozygote.	Aa looks like AA.
Incomplete dominance	Heterozygote is intermediate.	Blended F1 phenotype.
Codominance	Both alleles are expressed distinctly.	Both products are visible.


- A dominant allele does not subdue a recessive allele; dominance depends on how the phenotype is being measured.
- Alleles are just sequence variants in a gene.

For any character, dominance/recessiveness relationships of alleles depend on the level at which we examine the phenotype

⊕ It'll be explained better in the upcoming couple of slides

Tay Sach's features:
TAY SACHS

- Testing recommended
- Autosomal recessive
- Young death (<4 yrs.)
- Spot in macula (cherry red spots)
- Ashkenazi Jews
- CNS degeneration
- Hex A deficiency
- Storage disease




MENDELIAN GENETICS AND HUMANS
Human genetic disorders

Tay Sachs Disease

Inheritance Pattern:
-Autosomal recessive

Physical Effects:

- Nerve cells destroyed in brain and spinal cord
- Symptoms appear 3-6 months after birth
- Loss of motor control and atrophy of muscles, seizures
- Death



Hypotonia (decreased muscle tone)

#ADAM

Slide 9: Tay-Sachs clinical features and inheritance pattern.

Autosomal Recessive Disorders

- Tay-Sachs Disease
 - Usually occurs in Jewish people
 - Symptoms
 - Development slows at age 4 to 8 months
 - Neurological and Psychomotor impairment
 - Child gradually becomes blind and helpless, seizures, paralyzed, death by age 3 – 4 years old
 - Caused by gene on chromosome 15 → caused buildup of nonfunctional lysosomes in neurons
 - **Tay-Sachs disease** is fatal; a dysfunctional enzyme causes an accumulation of lipids in the brain

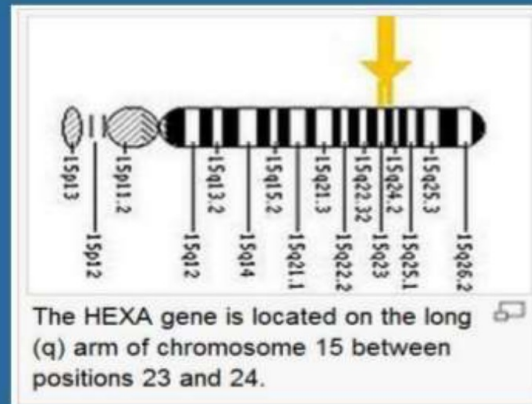
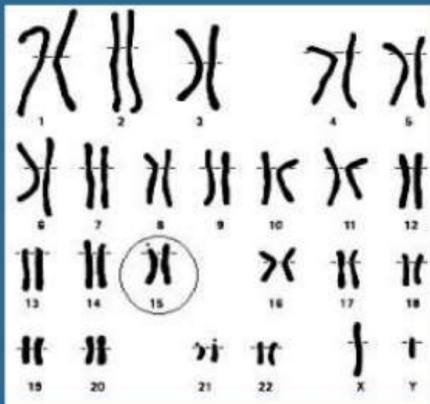
Slide 10: Tay-Sachs is an autosomal recessive disorder with early neurodegeneration.

3. Tay-Sachs disease: what the slides emphasize

- Caused by mutations on chromosome 15 in the HEXA gene, which leads to a lack of hexosaminidase A.
- The disease is especially associated with Ashkenazi Jews, and testing is recommended.
- Symptoms in the slides include developmental slowing, neurological and psychomotor impairment, blindness, seizures, paralysis, and death by about age 3-4 years.
- At the organismal level the allele behaves as recessive; at the biochemical level it is incompletely dominant; at the molecular level the alleles are codominant.

Causes of Tay-Sachs

The disease is caused by mutations on chromosome 15 in the HEXA gene, which produces a lack of hexosaminidase A.



Slide 8: the HEXA gene on chromosome 15 and loss of hexosaminidase A.

Dominant does not mean common

- Dominant alleles are not necessarily more common than recessive alleles.
- Polydactyly is the lecture example: the unusual trait is dominant, but the recessive five-digit allele is more prevalent in the population.

Frequency of Dominant Alleles

- Dominant alleles are not necessarily more common in populations than recessive alleles
- For example, Polydactyly one baby out of 400 in the United States is born with extra fingers or toes



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Slide 12: polydactyly shows that dominance and frequency are different.

- The allele for this unusual trait is dominant to the allele for the more common trait of five digits per appendage
- In this example, the recessive allele is far more prevalent than the population's dominant allele

Slide 13



4. Multiple alleles and pleiotropy





Multiple alleles

- Most genes exist in populations in more than two allelic forms.
- The ABO blood group has three alleles: I^A , I^B , and i .
- I^A adds the A carbohydrate, I^B adds the B carbohydrate, and i adds neither.
- Genotype to phenotype: $I^A I^A$ or $I^A i$ = A; $I^B I^B$ or $I^B i$ = B; $I^A I^B$ = AB; ii = O.

- The enzyme encoded by the I^A allele adds the A carbohydrate, whereas the enzyme encoded by the I^B allele adds the B carbohydrate; the enzyme encoded by the i allele adds neither
- enzyme(I)*

Figure 14.11

(a) The three alleles for the ABO blood groups and their carbohydrates				
Allele	I^A	I^B	i	
Carbohydrate	A 	B 	none	

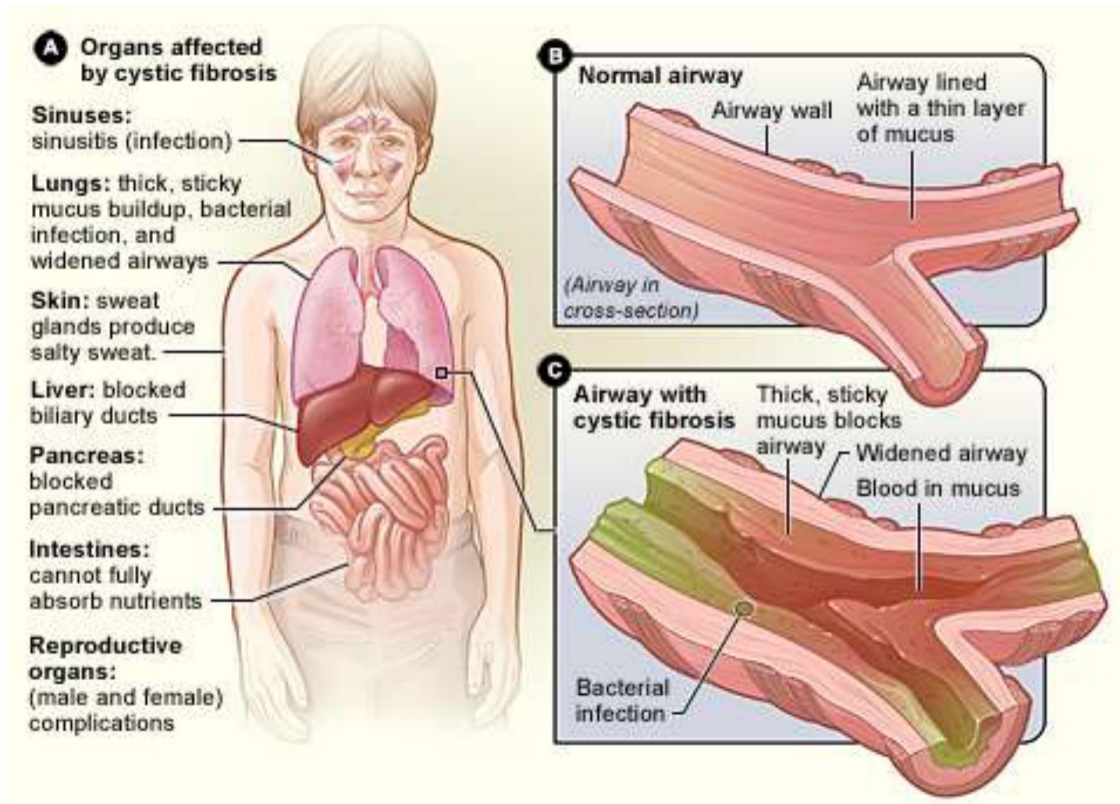
(b) Blood group genotypes and phenotypes				
Genotype	$I^A I^A$ or $I^A i$	$I^B I^B$ or $I^B i$	$I^A I^B$	ii
Red blood cell appearance				
Phenotype (blood group)	A	B	AB	O

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Slide 15: ABO alleles and blood-group phenotypes.

Pleiotropy

- Pleiotropy means one gene has multiple phenotypic effects.
- Cystic fibrosis and sickle-cell disease are the lecture examples of pleiotropic alleles.
- The cystic fibrosis diagram shows that many organs can be affected: sinuses, lungs, skin, liver, pancreas, intestines, and reproductive organs.



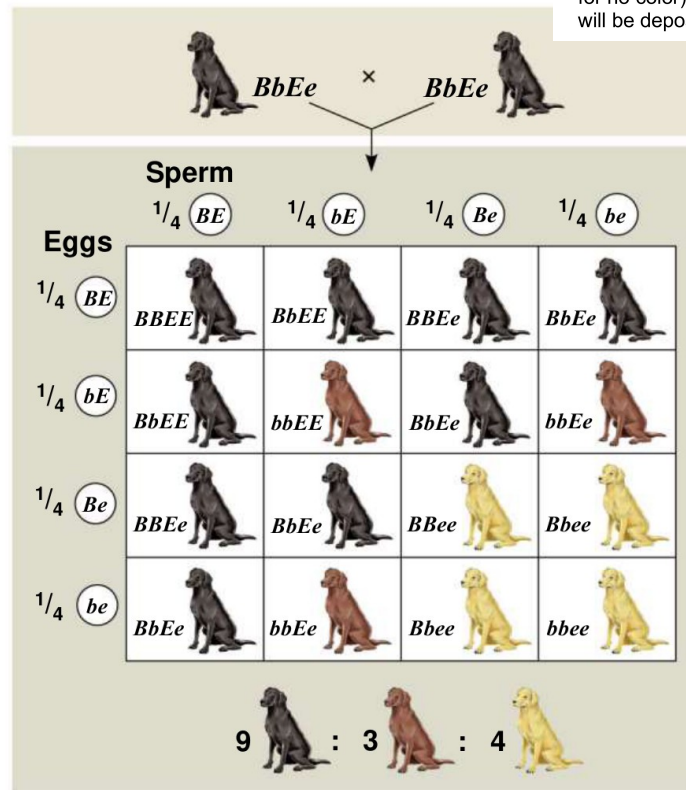
Slide 17: cystic fibrosis affects multiple organ systems.

5. Two or more genes can shape one trait

Epistasis

- Epistasis occurs when a gene at one locus alters the phenotypic expression of a gene at a second locus.
- The Labrador retriever example uses one gene for pigment color (B/b) and another for pigment deposition (C/c).
- The classic ratio shown in the slide is 9:3:4.

Figure 14.12



the Alleles (C/c) in the text are the same to alleles (E/e) in the diagram

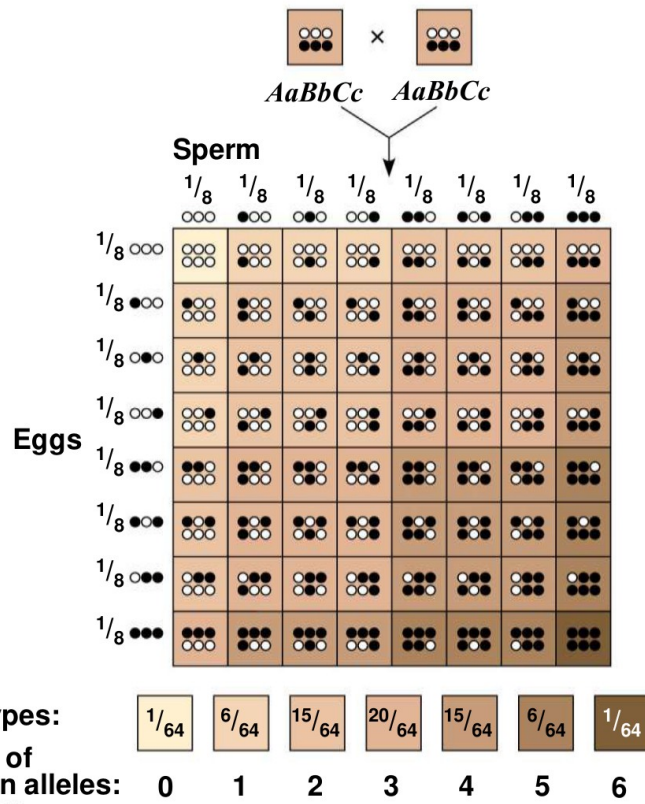
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Slide 20: Labrador coat color is the epistasis example.

Polygenic inheritance

- Quantitative characters vary along a continuum.
- This usually means polygenic inheritance: the additive effect of two or more genes on one phenotype.
- Human skin color is the slide example.
- More dark-skin alleles shift the phenotype toward darker pigmentation, producing a graded spectrum rather than neat categories.

Figure 14.13



Slide 22: additive alleles produce a smooth distribution of skin color.

Environment matters too

- Phenotype can depend on environment as well as genotype.
- The norm of reaction is the phenotypic range of a genotype influenced by the environment.
- Hydrangea flowers can be blue-violet or pink depending on soil acidity.
- Traits shaped by both genes and environment are called multifactorial.

Nature and Nurture: The Environmental Impact on Phenotype

- Another departure from Mendelian genetics arises when the phenotype for a character depends on environment as well as genotype
- The **norm of reaction** is the phenotypic range of a genotype influenced by the environment
- For example, hydrangea flowers of the same genotype range from blue-violet to pink, depending on soil acidity

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Slide 23: hydrangea color changes with the environment.

- Norms of reaction are generally broadest for polygenic characters
- Such characters are called **multifactorial** because genetic and environmental factors collectively influence phenotype

Slide 24

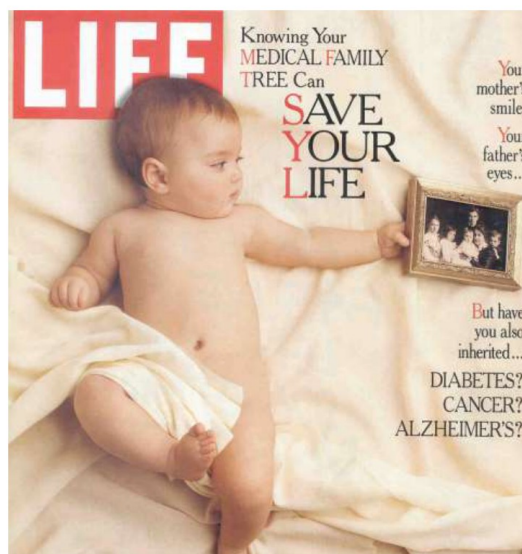
6. Mendelian genetics in humans

- Humans are not ideal research organisms because generation time is long, offspring numbers are small, and breeding experiments are unacceptable.
- Even so, Mendelian genetics remains the foundation of human genetics.

Pedigree analysis

- A pedigree is a family tree that shows the relationships of parents and children across generations.
- Pedigrees let you trace inheritance patterns and make predictions about future offspring.
- The multiplication and addition rules are used to calculate probabilities of specific phenotypes.
- A phenotype includes physical appearance, internal anatomy, physiology, and behavior.
- Phenotype reflects both genotype and environmental history.

Importance of Family History



- Understanding the past is the key to predicting the future.

Slides 27-28: pedigrees plus probability let you predict inheritance.

Integrating a Mendelian View of Heredity and Variation

- An organism's phenotype includes its physical appearance, internal anatomy, physiology, and behavior
- An organism's phenotype reflects its overall genotype and unique environmental history

slide 25

7. Ultra-quick comparison table

Concept	One-line meaning	Example from slides
Incomplete dominance	Heterozygote is intermediate.	Tay-Sachs enzyme activity level
Codominance	Both alleles are seen separately.	ABO blood group
Multiple alleles	More than two allele forms exist in a population.	ABO system
Pleiotropy	One gene affects several traits.	Cystic fibrosis
Epistasis	One gene masks or modifies another gene.	Labrador coat color
Polygenic inheritance	Many genes add up to one phenotype.	Skin color
Multifactorial trait	Genes plus environment shape the phenotype.	Hydrangea flowers

يا ربِّ لك الحمد كما ينبغي لجلال وجهك و عظيم سلطانك، اللهم ربِّ لك الحمد حتى ترضى، ولك الحمد ربِّ إذا رضيت، ولك الحمد ربِّ بعد الرضا

Thanks for using this sheet.

If you found it helpful, please share it.

8. Slide-by-slide coverage map

Slide	Key point
1	Title slide: Single-Gene Inheritance
2	Importance of family history
3	Lecture objectives
4	Inheritance patterns are often more complex than simple Mendelian genetics
5	Ways single-gene inheritance can deviate from simple Mendelian patterns
6	Degrees of dominance
7	Dominance is not an interaction where one allele subdues another
8	Tay-Sachs cause: HEXA mutation on chromosome 15
9	Tay-Sachs features
10	Tay-Sachs as an autosomal recessive disorder with neurological decline
11	Tay-Sachs behaves differently at organismal, biochemical, and molecular levels
12	Dominant alleles are not necessarily more common
13	Polydactyly example
14	Multiple alleles in general
15	ABO blood group genotypes and phenotypes
16	Pleiotropy definition
17	Cystic fibrosis illustration
18	Traits determined by two or more genes
19	Epistasis definition and Labrador example setup
20	Epistasis figure and 9:3:4 ratio
21	Polygenic inheritance definition
22	Skin color polygenic figure
23	Environment impacts phenotype; norm of reaction
24	Broad norms of reaction for polygenic characters; multifactorial traits
25	Integrating heredity and variation
26	Many human traits follow Mendelian patterns
27	Pedigree analysis definition
28	Pedigrees and probability rules