

Credit: Patinya (Adobe Stock)

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- Gregor Johann Mendel (Austrian), 1865
  - Set the framework for genetics
  - Conducted methodical, quantitative analyses using large sample sizes
  - Because of Mendel's work, the fundamental principles of heredity were revealed



Gregor Johann Mendel (1822–1884).  
(Courtesy of Professor William Bateson,  
London.)

Portrait of Gregor Johann Mendel - Garrison.  
[Wellcome Collection](https://wellcomecollection.org/works/tc5xq5ad).  
[wellcomecollection.org/works/tc5xq5ad](https://wellcomecollection.org/works/tc5xq5ad)  
(CC BY 4.0)

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<https://youtu.be/PeSSI2iHrWE>

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- Physical characteristics are expressed through genes carried on chromosomes.
- Each pair of homologous chromosomes has the same linear order of genes.
- The two genetic copies may or may not encode the same version of a characteristic.
- Gene variants at the same relative locations on homologous chromosomes are called **alleles**.

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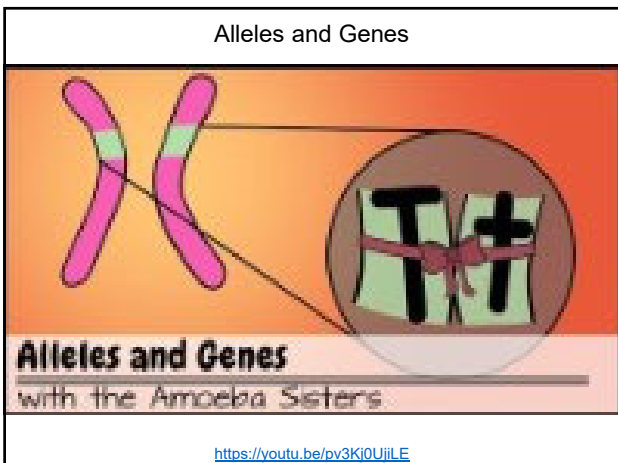
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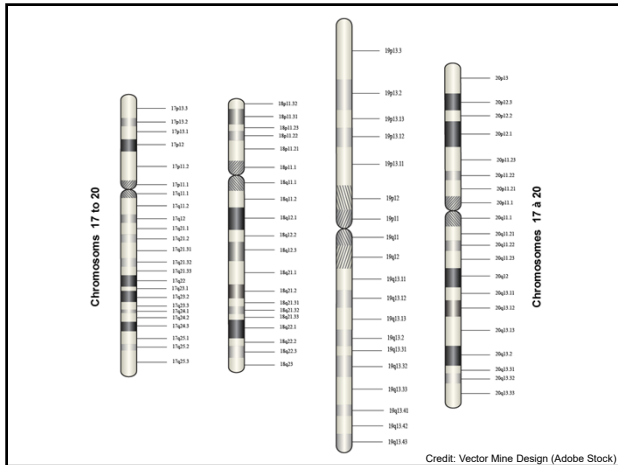
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## Phenotypes and Genotypes

- Two alleles for a given gene in a diploid organism are expressed and interact to produce physical characteristics.
- The **observable traits** expressed by an organism are referred to as its **phenotype**.
- The **underlying genetic makeup**, consisting of both physically visible and non-expressed alleles, is called its **genotype**.

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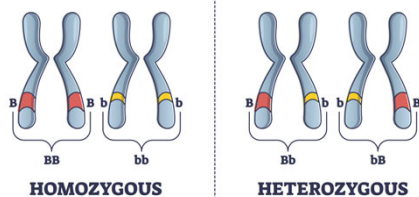
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- If the two alleles are the **same** the organism is said to be **homozygous** for the trait.
- If the two alleles are **different** the organism is said to be **heterozygous** for the trait.



Credit: VectorMine (Adobe Stock)

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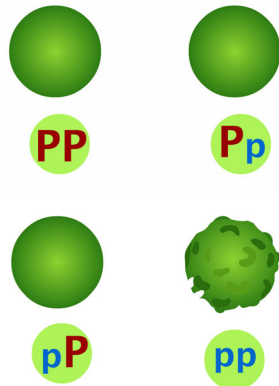
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- A heterozygous organism will appear the same as some homozygous organisms.

- This implies that one of the genes is **dominant** over the other (**recessive**).

- Homozygous dominant and heterozygous organisms will look identical.



Credit: LuckySoul (Adobe Stock)

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- Several conventions exist for referring to genes and alleles:

- A letter is used to represent the trait.
  - Uppercase for dominant.
  - Lowercase for recessive.
- The letter is usually italicized.

Example:

V for violet flowers	VV – violet
v for white flowers	Vv – violet
	vv – white

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## Punnett Squares

- Reginald Punnett (British)
  - Developed a chart that allows you to easily determine the expected percentage of different genotypes in the offspring of two parents. (Punnett Square)



Reginald Punnett  
Non-commercial / Non-commercial presentation or lecture  
One-off use in an educational establishment, including use in a non-commercial presentation or lecture provided it is made at no cost to the end user  
Name of educational establishment: College Sturgeon Heights Collegiate

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## Monohybrids and the Punnett Square Guinea Pigs

Genetics Terms - video 1

**Monohybrid Crosses**

with the Amazing Guinea Pigs

<https://youtu.be/i-0rSv6oxSY>

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CSH Cold Spring Harbor Laboratory  
DNA LEARNING CENTER

**: Genetic inheritance follows rules.**

I'm Reginald Punnett, William Bateson and I were very keen on Mendel's laws of inheritance.

JUMP TO

DESCRIPTION TRANSCRIPT KEYWORDS INFO

Reginald Punnett and William Bateson explain Mendel's ratios.

Source: <https://www.dnalc.org/view/16192-Animation-5-Genetic-inheritance-follows-rules-.html>

<http://www.dnalc.org/view/16192-Animation-5-Genetic-inheritance-follows-rules-.html>

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Y = yellow ●  
y = green ●

Parent 1  
Yy ●

Parent 2  
Yy ●

	Y	y
Y	YY	Yy
y	Yy	yy

YY = yellow ●  
2 Yy = yellow ●  
yy = green ●

$\frac{1}{4} = 25\% \text{ YY}$   $\frac{2}{4} = 50\% \text{ Yy}$   $\frac{1}{4} = 25\% \text{ yy}$   
 $\frac{3}{4} = 75\% \text{ yellow } \bullet$   $\frac{1}{4} = 25\% \text{ green } \bullet$

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## Example

- Use a Punnett square to predict the offspring in a cross between a dwarf pea plant (homozygous recessive) and a tall pea plant (heterozygous).
- What is the phenotypic ratio of the offspring?

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T = tall  
t = dwarf

Parent 1 = tt  
Parent 2 = Tt

	t	t
T	Tt	Tt
t	tt	tt

$$Tt = \text{tall} = \frac{2}{4} = 50\%$$

$$tt = \text{dwarf} = \frac{2}{4} = 50\%$$

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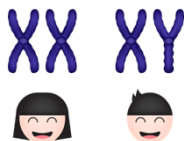
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## Sex Chromosomes

- Males have one X and one Y chromosome.
- Females have two X chromosomes.
  - One of the X chromosomes in females is turned off.
  - The X chromosome is about three times bigger than the Y chromosome.



Credit: olando (Adobe Stock)

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## Sex-Linked Traits

- Genes located on the sex chromosomes are called sex-linked genes.
  - The traits are thus called sex-linked traits.
- Most sex-linked genes are on the X chromosome.
  - These genes (and associated traits) are often called X-linked.

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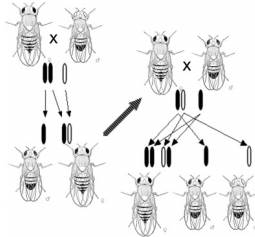
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- Thomas Hunt Morgan (American), 1910
  - First identified the existence of X-linked traits.
    - eye color in *Drosophila*



Thomas Hunt Morgan - U.S. National Library of Medicine (public domain)

White mutation and sex-linked inheritance. The Physical Basis of Heredity. Thomas Hunt Morgan. Philadelphia: J.B. Lippincott Company. 1919 (public domain)

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$X^W$  = red eyes  
 $X^w$  = white eyes

male  
 $X^wY$  (white eyes)

female $X^W X^w$ (red eyes)	$X^W$	$X^W X^w$	$X^W Y$
	$X^w$	$X^w X^w$	$X^w Y$

25% female  $X^W X^w$  (red eyes)  
25% female  $X^w X^w$  (white eyes)  
25% male  $X^W Y$  (red eyes)  
25% male  $X^w Y$  (white eyes)




Image: Kim (CC BY-NC-ND 2.0)

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$X^W$  = red eyes  
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male  
 $X^W Y$  (red eyes)

female $X^W X^w$ (red eyes)	$X^W$	$X^W X^W$	$X^W Y$
	$X^w$	$X^W X^w$	$X^w Y$

25% female  $X^W X^W$  (red eyes)  
25% female  $X^W X^w$  (red eyes)  
25% male  $X^W Y$  (red eyes)  
25% male  $X^w Y$  (white eyes)




Image: Kim (CC BY-NC-ND 2.0)

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### Human Sex-linked Disorders

- Because human males need to inherit only one recessive X allele to be affected, X-linked disorders are disproportionately observed in males.
- When females inherit one recessive X-linked mutant allele and one dominant X-linked wild-type allele, they are carriers of the trait and are typically unaffected.

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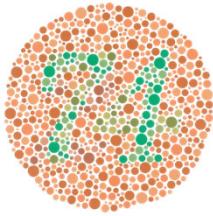
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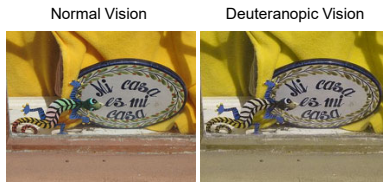


## Some Examples

- Red-Green Color Blindness
  - Unable to tell the difference between red and green



Ishihara color test plate - Shinobu Ishihara (public domain)



Images: Ottmar Liebert ([CC BY-SA 2.0](https://creativecommons.org/licenses/by-sa/2.0/))

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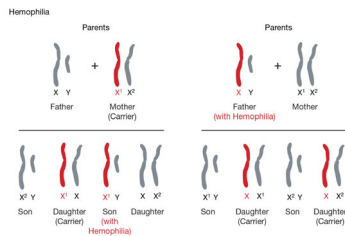
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- Hemophilia
  - Hemophilia is a rare disorder in which your blood doesn't clot normally because it lacks sufficient blood-clotting proteins (clotting factors).



Courtesy: National Human Genome Research Institute ([genome.gov](https://www.genome.gov/))

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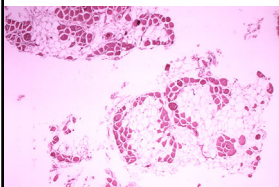
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- Duchenne muscular dystrophy (DMD)
  - Affects the muscles, leading to muscle wasting that gets worse over time
  - Arises from mutations of the DMD gene located on the X chromosome
  - Becker muscular dystrophy, a milder form of muscular dystrophy, is also caused by DNA variants in the DMD gene.



This photomicrograph revealed histopathologic changes found in a skeletal muscle tissue harvested from the gastrocnemius muscle, of a patient with a fatal case of Duchenne muscular dystrophy (DMD). This cross section of the muscle, shows extensive replacement of muscle fibers, by adipose, or fat cells.

Image: CDC/ Dr. Edwin P. Ewing Jr. (Public Domain)

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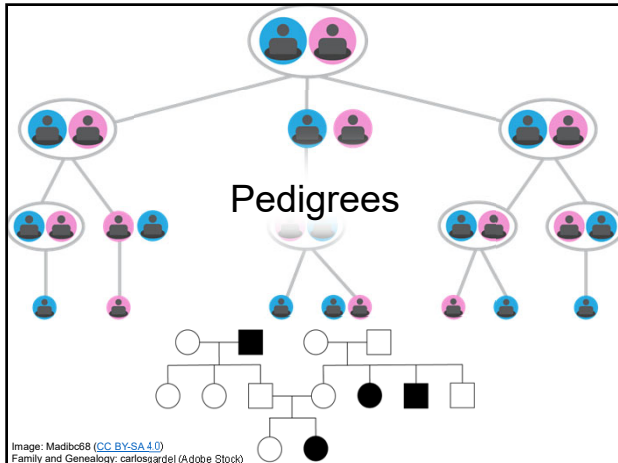
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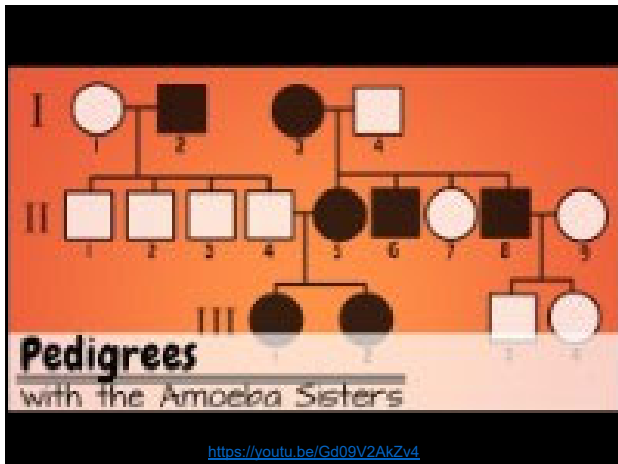
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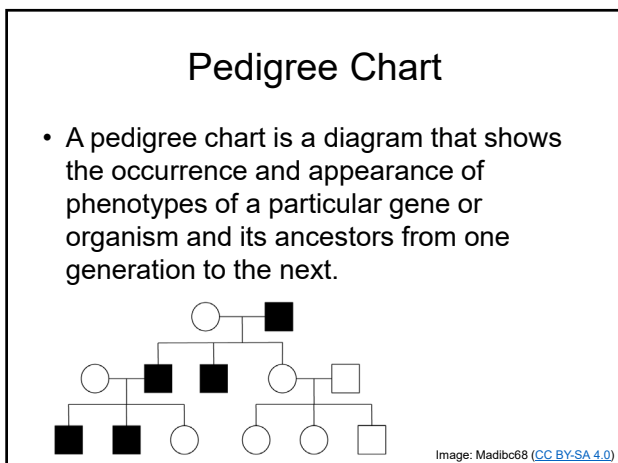
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- Pedigrees use a standardized set of symbols:
  - Squares represent males.  $\square$
  - Circles represent females.  $\circ$
  - If the sex of the person is unknown a diamond is used.  $\diamond$
  - Someone with the phenotype in question is represented by a filled-in symbol.  $\blacksquare$
  - Heterozygotes (carriers) are indicated by a dot inside a symbol or a half-filled symbol.  $\square\cdot$
  - Generations are indicated by Roman numerals (I,II,III).

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The filled symbols represent the affected individuals. You may assume that the affected allele is rare and therefore individuals marrying into the family do not have it.

1. State the genotype for the individuals labelled 1-5 using the letter "A".
2. If individuals 2 and 3 were to have another son, what are the chances that he will be affected? 50%

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