

when	who	what
1865	Gregor Mendel	<i>Experiments on Plant Hybridization</i>
1910	Thomas Hunt Morgan	Chromosomes are carriers of genes
1941	E. L. Tatum and G. W. Beadle	Genes encode proteins
1953	J. D. Watson and F. Crick	DNA double helix structure
1961	Marshall W. Nirenberg	Genetic Code discovery
1968	M. Meselson and R. Yuan	Restriction Enzyme discovery
1996	Roslin Institute	Cloned Sheep

## Mendel's Laws:

**Law of Segregation:** There are two alleles for each gene; during the gamete formation, the two alleles of one gene segregate from each other independently

**Law of Independent Assortment:** During the gamete formation, genes from different chromosomes assort independently and combine randomly



Complete penetrance and unvarying expressivity



Incomplete penetrance and unvarying expressivity



Complete penetrance and variable expressivity



Incomplete penetrance and variable expressivity

**X-linked Recessive Traits:**  
Appear more often in males.  
Not passed from father to son  
Passed from carrier mother to son  
Skip generations

**X-linked Dominant Traits:**  
Appear in both males and females.  
Affected males must have affected mother  
Affected males pass the trait to all of their daughters  
Do not skip generations

**Y-linked Traits:**  
Appear only in males  
Pass from father to son  
Do not skip generations

## Linked Genes

Parents > Recombinants (RF < 50%)

Linked genes must be syntenic and sufficiently close together on the same chromosome so that they do not assort independently.

## Unlinked Genes

Parents = Recombinants (RF = 50%)

Occurs either when two genes are on different chromosomes or when they are sufficiently far apart on the same chromosome that at least one crossover occurs between them in every meiosis.

Phase (Mitosis)	# Chromosomes	# Chromatids
Prophase	46	92
Metaphase	46	92
Anaphase	92	92
Telophase	92	92
End of Mitosis (separated cells)	46	46

Phase (Meiosis I)	# Chromosomes	# Chromatids
Prophase I	46	92
Metaphase I	46	92
Anaphase I	46	92
Telophase I	46	92
End of Meiosis I (separated cells)	23	46

Phase (Meiosis II)	# Chromosomes	# Chromatids
Prophase II	23	46
Metaphase II	23	46
Anaphase II	46	46
Telophase II	46	46
End of Meiosis II (separated cells)	23	23

**-Hardy Weinberg Equation:**  $p^2 + 2pq + q^2 = 1$ , Heterozygosity(H) =  $2pq \rightarrow$  and  $P'_m = P_t$ ,  $P'_t = P_m + P_t/2$  when  $P_t = P_m = P$  population is in HWE

**- Inbreeding coefficient F:**  $F = \frac{E(F(Aa)) - O(F(Aa))}{E(F(Aa))} = \frac{E(H) - O(H)}{E(H)} =$

Inbreeding increases homozygosity, but relative allele frequencies remain constant if:  $F=1 \rightarrow$  HWE, if  $0 < F < 1 \rightarrow$  inbreeding, if  $1 < F < 0 \rightarrow$  outbreeding.

**-Chi-Squared:**  $\chi^2 = \sum \frac{(obs-exp)^2}{exp}$ , if  $\chi^2 > 5\%$  in HWE if < not in HWE

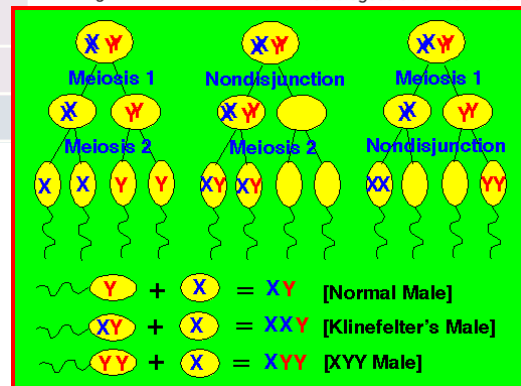
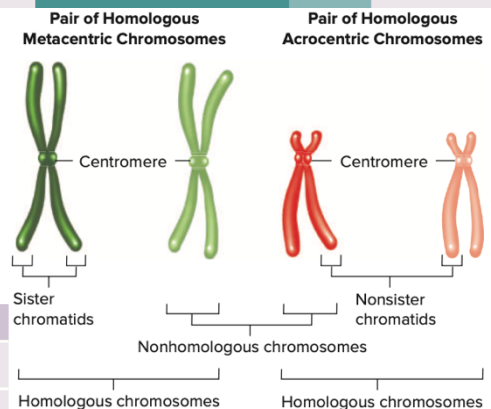
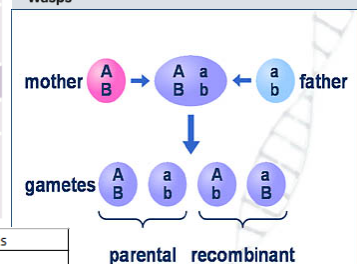
**-Genetic drift:**  $P(\Delta 1 = 0 - 2N) = \left( \frac{(2N)!}{k!(2N-k)!} \right) p^k q^{2N-k} \rightarrow$  calculate for all  $\Delta 1$  then  $P(\text{fixed} \rightarrow 1G) = P(\Delta 1=0) + P(\Delta 1=2N)$  and  $P(\text{change}) = 1 - P(\Delta 1=N)$

Mutation Rate:  $\mu = \frac{\text{Number } A1 \rightarrow A2}{\text{Number of bases} \times \text{years}} = \frac{\text{Number } A1 \rightarrow A2}{\text{total } A \text{ loci}}$

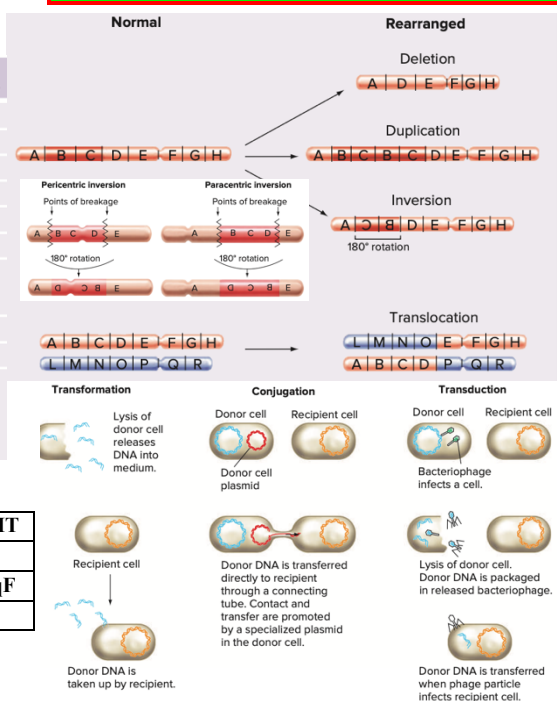
Gene Interaction	Example	F <sub>2</sub> Genotypic Ratios from an F <sub>1</sub> Dihybrid Cross				F <sub>2</sub> Phenotypic Ratio
		A- B-	A- bb	aa B-	aa bb	
<b>Additive:</b> Four distinct F <sub>2</sub> phenotypes	Lentil: seed coat color (see Fig. 3.10a)	9	3	3	1	9:3:3:1
<b>Recessive epistasis:</b> When homozygous, recessive allele of one gene masks both alleles of another gene	Labrador retriever: coat color (see Fig. 3.12b)	9	3	3	1	9:3:4
<b>Reciprocal recessive epistasis:</b> When homozygous, recessive allele of each gene masks the dominant allele of the other gene	Sweet pea: flower color (see Fig. 3.15b)	9	3	3	1	9:7
<b>Dominant epistasis I:</b> Dominant allele of one gene hides effects of both alleles of the other gene	Summer squash: color (see Fig. 3.17a)	9	3	3	1	12:3:1
<b>Dominant epistasis II:</b> Dominant allele of one gene hides effects of dominant allele of other gene	Chicken feathers: color (see Fig. 3.18a)	9	3	3	1	13:3
<b>Redundancy:</b> Only one dominant allele of either of two genes is necessary to produce phenotype	Maize: leaf development (see Fig. 3.19b)	9	3	3	1	15:1

Type of Dominance	A <sup>1</sup> A <sup>1</sup>	A <sup>2</sup> A <sup>2</sup>	A <sup>1</sup> A <sup>2</sup> hybrids
Complete			A <sup>1</sup> is dominant to A <sup>2</sup> A <sup>2</sup> is recessive to A <sup>1</sup>
Complete			A <sup>2</sup> is dominant to A <sup>1</sup> A <sup>1</sup> is recessive to A <sup>2</sup>
Incomplete			A <sup>1</sup> and A <sup>2</sup> are incompletely dominant relative to each other
Codominant			A <sup>1</sup> and A <sup>2</sup> are codominant relative to each other

	♀	♂
<b>Humans and Drosophila</b>	XX	XY
<b>Moths and C. elegans</b>	XX (hermaphrodites in C. elegans)	XO
<b>Birds and Butterflies</b>	ZW	ZZ
<b>Bees and Wasps</b>	Diploid	Haploid



Chromosomes	Syndrome
<b>Autosomes</b>	
Trisomic 21	Down
Trisomic 13	Patau
Trisomic 18	Edwards
<b>Sex chromosomes, females</b>	
XO, monosomic	Turner
XXX, trisomic	
XXXX, tetrasomic	
XXXXX, pentasomic	
<b>Sex chromosomes, males</b>	
XY, trisomic	Normal
XXY, trisomic	
XXYY, tetrasomic	
XXXY, tetrasomic	
XXXXY, pentasomic	
XXXXXY, hexasomic	Klinefelter



	HWE	WRIGHT
F(A <sub>1</sub> A <sub>1</sub> )	p <sup>2</sup>	p <sup>2</sup> +pqF
F(A <sub>1</sub> A <sub>2</sub> )	2pq	2pq-2pqF
F(A <sub>2</sub> A <sub>2</sub> )	q <sup>2</sup>	q <sup>2</sup> +pqF

## LOD Ratio

R, N = # recombinants, # non-recomb

$\Theta = R / R+N$

$OR = (\Theta(\Theta-1)^N) / (0.5)^{N+R}$ , LOD = Log<sub>10</sub>OR

If LOD ≥ N → Associated

If LOD < N → Not associated