Inheritance

Today's Outline

- Gregor Mendel
- Theory of segregation
- Theory of independent assortment
- Sources of variation in populations
- Chromosomal basis of inheritance
- Human genetics & ethics

Gregor Johann Mendel

Gregor Johann Mendel (1822-1884)

Treatise on Plant Hybrids (1865)

Darwin & Mendel – near miss

Darwin – (1809-1882)
Mendel – (1822-1884)

Darwin – Origin of species (1859)
Mendel - Treatise on Plant Hybrids (1865)

Modern Synthesis

The unification of Mendel's and Darwin's theories by geneticists, paleontologists and evolutionary biologists (1920-1947).

More than 50 years after Darwin's and Mendel's publications.

Terms

Gene
Locus
Allele
Diploid
Dominant
Recessive
Homozygous
Heterozygous
Genotype
phenotype
Crosses
Punnett-square
Segregation
Plieiotropy
Variation
Autosomes
Linkage
Duplication
Inversion
Deletion
translocation

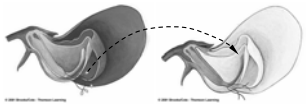
Mendel's Experiments

The Garden Pea Plant

Self-pollinating

True breeding (different alleles not normally introduced)

Can be experimentally cross-pollinated



Mendel's Experiments

Tracking Generations

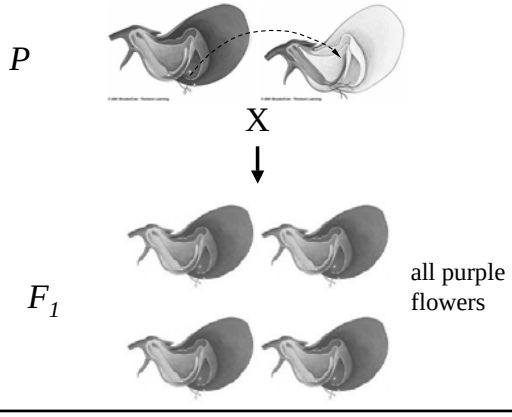
Parental generation mates to produce — *P*

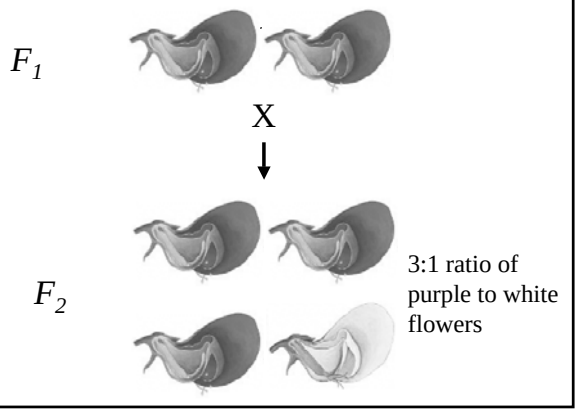
↓

First-generation offspring mate to produce — F_1

↓

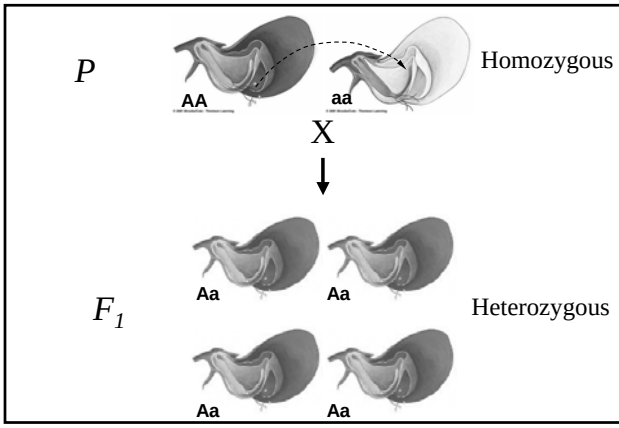
Second-generation offspring — F_2

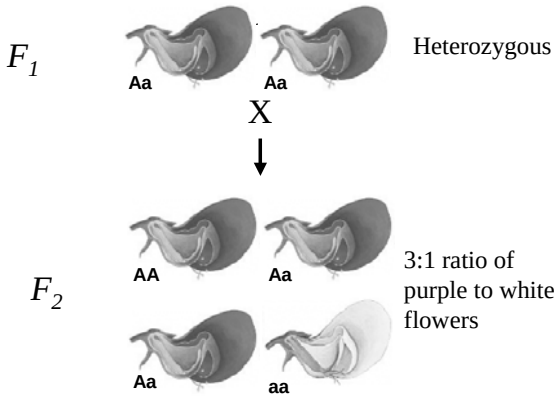




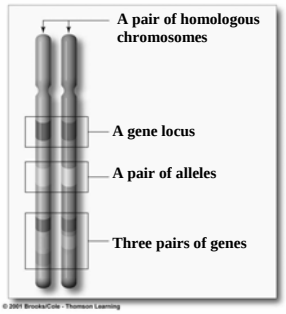
Mendel's hypothesis

1. Each Parental (*P*) pea plant contributed 1 of 2 units (alleles) to the hybrid offspring.
2. The purple flowered pea plant had two dominant units while the white flower pea plant had two recessive units (both are homozygous).



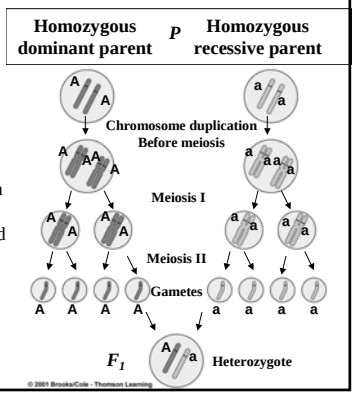


Genetic Terms

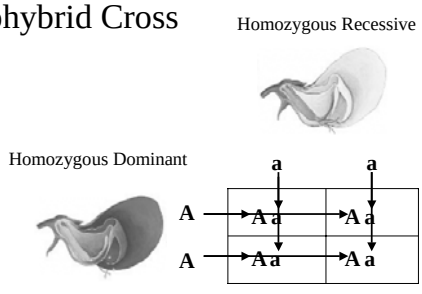


Mendel's Theory of Segregation

Diploid cells have pairs of genes on homologous Chromosomes. The two genes of each pair are separated from each other during meiosis, so They end up in different gametes.



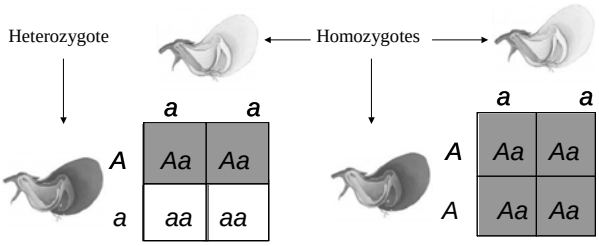
Punnett-Square Method Monohybrid Cross



Phenotype & Genotype


Phenotype	Purple flower	White flower
Genotype	AA or Aa	aa

Punnett Squares of Test Crosses

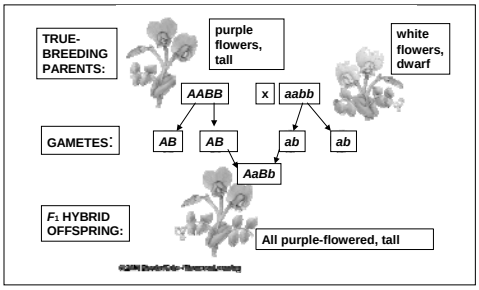


Dihybrid Cross

Crosses designed to evaluate two characteristics.

For example: Mendel performed crosses between purple flowered plants and long stems with white flowered and short stem.

Dihybrid Cross



Dihybrid Heterozygotes & Theory of Independent Assortment

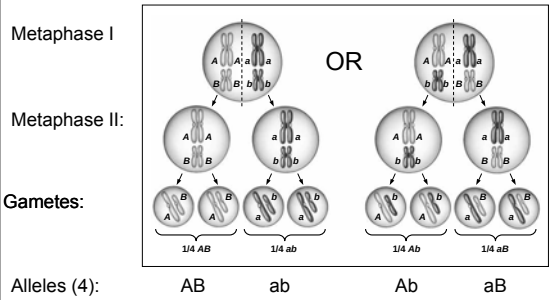
How would the gametes of these dihybrid heterozygotes assort during meiosis?

Based on his dihybrid crosses, Mendel proposed that alleles will assort independently from one another.

Of course, this is dependent on whether the genes are on the same or different chromosomes.

- If the genes are on different chromosomes they will assort independently.
- If they are on the same chromosome they will be linked and assort together.

Independent Assortment

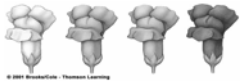


Incomplete Dominance
Flower Color in Snapdragons

Pink-flowered plant (heterozygote) X Pink-flowered plant (heterozygote)

↓

White-, pink-, and red-flowered plants in a 1:2:1 ratio



aa Aa Aa AA

Codominance
ABO Blood Types

Blood Type	Genotype
A	AA or AO
B	BB or BO
AB	AB
O	OO

A and B alleles are codominant. O allele is recessive.

Codominance
ABO Blood Types

Cross a Father with Blood Type A?
and a Mother with Blood Type AB:

Possible Outcome No. 1

	A	B
A		
A		

Possible Outcome No. 2

	A	B
A		
O		

Codominance
ABO Blood Types

Cross a Father with Blood Type A?
and a Mother with Blood Type AB:

Possible Outcome No. 1

	A	B
A	AA	AB
A	AA	AB

Possible Outcome No. 2

	A	B
A		
O		

Codominance
ABO Blood Types

Cross a Father with Blood Type A?
and a Mother with Blood Type AB:

Possible Outcome No. 1

	A	B
A	AA	AB
A	AA	AB

Possible Outcome No. 2

	A	B
A	AA	AB
O	AO	BO

Pleiotropy

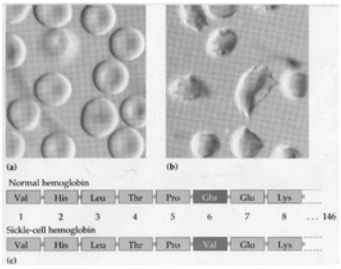
pleiotropy - genes affect several different characters in the same individual.

Example: same gene affects fur color, cross-eyedness, and vocalizations in Siamese cats

Pleiotropy

Sickle-cell Anemia

$Hb^A\ Hb^A$ $Hb^A\ Hb^S$ $Hb^S\ Hb^S$



Epistasis: Interactions Between Gene Pairs

F_1: BBEE x bbee → BbEe
Black x Yellow

F_2: BbEe x BbEe

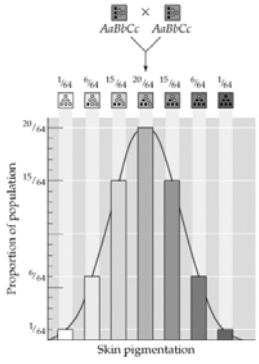
	BE	Be	bE	be
BE	BBEE	BBEe	BbEE	BbEe
Be	BBEe	BBee	BbEe	Bbee
bE	BbEE	BbEe	bbEE	bbEe
be	BbEe	Bbee	bbEe	bbee

B–black
b–brown
E–melanin deposits
e–no melanin deposits

Polygenic Traits

Some phenotypic characters are determined by more than one gene.

Example: Human skin color



Other Influences on Gene Expression

Environment

Causes variation in how genotype is translated into phenotype.

Mutation

Produces new alleles.

Size at maturity depends on environment

platyfish

Genotype 1: Low food — High food

Genotype 2: Low food — High food

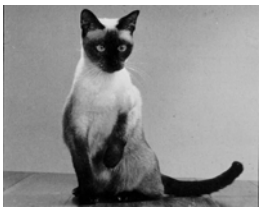
Genotype 3: Low food — High food

Small — **Size at Maturity** — **Large**

Environmental Effects on Phenotype

Light coat color

Warm environment



Dark coat color

Cold environment

Sources of Variation

Sexual reproduction allows offspring to have both different genotypes and different phenotypes than the parents. Sexual reproduction is an important source of genetic variation.

Independent assortment of chromosomes during meiosis produces gametes with new combinations.

Chromosomal crossing-over during meiosis produces new combinations of linked genes in homologous chromosomes.

The random fusion of gametes from both parents produces additional variation.

Sexual reproduction provides diversity for a species to survive environmental Change.

Chromosomes

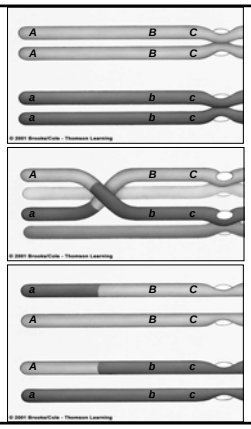
Autosomal – all chromosomes except sex chromosomes. Same for men and women.

Sex chromosomes – X and Y Chromosomes. They are nonidentical homologues.

- Women have 2 X chromosomes
- Men have one each of X and Y chromosomes.

Chromosomal Alterations

1. Crossover
2. Duplication
3. Inversion
4. Deletion
5. Translocation



Crossover during prophase I of Meiosis.

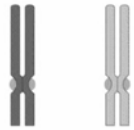


Fig. 11.2, p. 168

Duplication

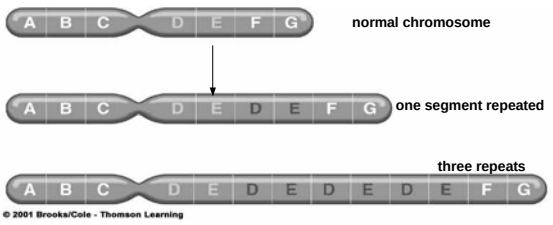
Gene sequence that is repeated several to hundreds of times

Duplications occur in normal chromosomes

May have adaptive advantage

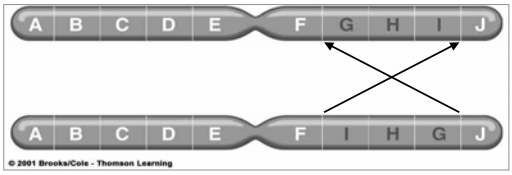
Useful mutations may occur in copy

Duplication



Inversion

A linear stretch of DNA is reversed within the chromosome

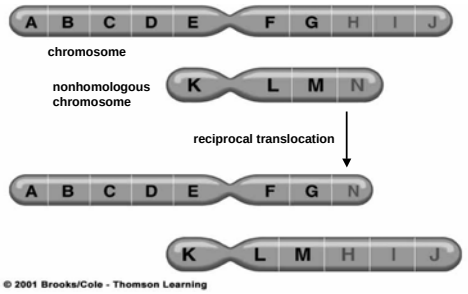


Translocation

A piece of one chromosome becomes attached to another nonhomologous chromosome

Most are reciprocal

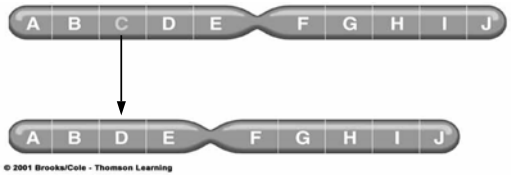
Translocation



Deletion

Loss of some segment of a chromosome

Most are lethal or cause serious disorder



Aneuploidy

Individuals have one extra or less chromosome ($2n + 1$ or $2n - 1$)

Major cause of human reproductive failure

Most human miscarriages are aneuploids

Polyploidy

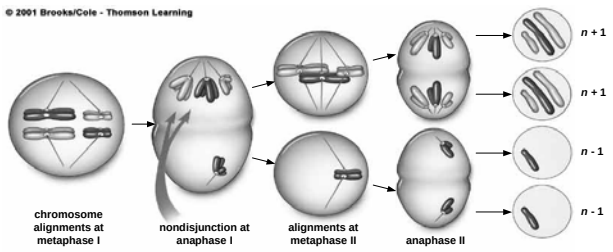
Individuals have three or more of each type of chromosome ($3n$, $4n$)

Common in flowering plants

Lethal for humans

- 99% die before birth
- Newborns die soon after birth

Nondisjunction



Down Syndrome

- Trisomy of chromosome 21
- Mental impairment and a variety of additional defects
- Can be detected before birth
- Risk of Down syndrome increases dramatically in mothers over age 35

Turner Syndrome

- Inheritance of only one X (XO)
- 98% spontaneously aborted
- Survivors are short, infertile females
  - No functional ovaries
  - Secondary sexual traits reduced
  - May be treated with hormones, surgery

Klinefelter Syndrome

- XXY condition
- Results mainly from nondisjunction in mother (67%)
- Phenotype is tall males
  - Sterile or nearly so
  - Feminized traits (sparse facial hair, somewhat enlarged breasts)
  - Treated with testosterone injections

XYY Condition

- Taller than average males
- Most otherwise phenotypically normal
- Some mentally impaired
- Once thought to be predisposed to criminal behavior, but studies now discredit