

BIOL 230: Cell & Molecular Biology

Fall 2019

17-205

W, Nov. 20

<http://accounts.smccd.edu/staplesn/biol230/>

1. Pre-Lab writeups due each Mon. (for both M&W!!) **WRITTEN!!!**
briefly, **What? Why? How?** for each expt. Question & **Hypothesis?!**
2. **GENETICS PRIMER:** http://anthro.palomar.edu/mendel/mendel_1.htm
3. **Prelab: do Mendelian Genetics exercises (A-C for Wed., 11/20) –**
& Ch. 12 Homework Problems:
 - a) <http://sonic.net/~nbs/projects/anthro201/>; *** <http://sonic.net/~nbs/projects/anthro201/exper/>
 - b) http://www.biology.arizona.edu/mendelian_genetics/mendelian_genetics.html ***
 - c) <https://www.ndsu.edu/pubweb/~mcclean/plsc431/mendel/mendel1.htm> (see left side INDEX!)
> <https://www.ndsu.edu/pubweb/~mcclean/plsc431/mendel/mendel9.htm> (see left side INDEX!)
 - d) <https://concord.org/teaching-genetics/dragons/> DRAGONS!!! – see Geniverse & Genigames
 - e) <http://www.dnafb.org/> → Classical Genetics, and Genetic Organization.
 - f) **TURN IN** your calculations of your D1S80 Genotype!! Also: **ARE** you an Alien Clone????
4. **LAB THIS week: Genotypes and Phenotypes!! PROBABILITIES!!!**
5. **Find Anastasia & Alien GEL DATA under ADDITIONAL MATERIALS.**
6. **Extra Credit: STEM SPEAKER SERIES, ALL DUE by THIS WEEK!!!!**
7. ******NOTEBOOK/Lab MANUAL is due TODAY!!**
8. **RESEARCH Outlines will be reviewed by tonight!!! FINAL REPORT due week of Wed., Dec. 4th. NO EXCEPTIONS!!!**

REVIEW

1. **Ch. 11:** Describe and Diagram the **4 phases of the cell cycle**, and how they are regulated by **Cyclin/CDK complexes**.
2. Diagram and compare the 4 main phases of **Mitosis**, **Meiosis I** & **Meiosis II**.
 - Chromosome, Chromatid, Centromere, Centriole, Spindle, Cortical MT's, Spindle fibers/ MT's, Sister Chromatid, Homologous Chromosome, Nuclear Envelope, Cytokinesis (plants/animals).

TODAY's Objectives: Students should be able to

1. Describe and diagram how **meiosis** generates **diversity** in gametes.
2. Define and explain the function of **Apoptosis**.
3. Describe **Mendel's experiments**, their results, and how these lead him to formulate the Laws of **Segregation** and **Independent Assortment**.
 - *His methods, choice of organism, choice of characters, Monohybrid & Dihybrid Crosses.*
4. Define and give examples of **gene**, **allele**, **dominant**, **recessive**, **homozygote**, **heterozygote**, **Genotype**, **Phenotype**, **monohybrid**, **dihybrid**, **true-breeding/purebred**, and **locus**.

❖ **LAST Objectives & Study Guide Questions are your HOMEWORK between classes!!! DUE NEXT WED. at the END of LECTURE!!**

11.6) Reproduction: Sexual & Asexual

- The cell cycle can repeat itself many times, forming a clone of genetically identical cells.
- **Asexual** reproduction produces an organism genetically identical to the parent.
 - Any genetic variety is the result of mutations.
- In **Sexual** reproduction,
 - two haploid gametes (**n # chromosomes**)
 - (one from each parent)
 - unite in fertilization to form a genetically unique, diploid zygote (**2n # chromosomes**)

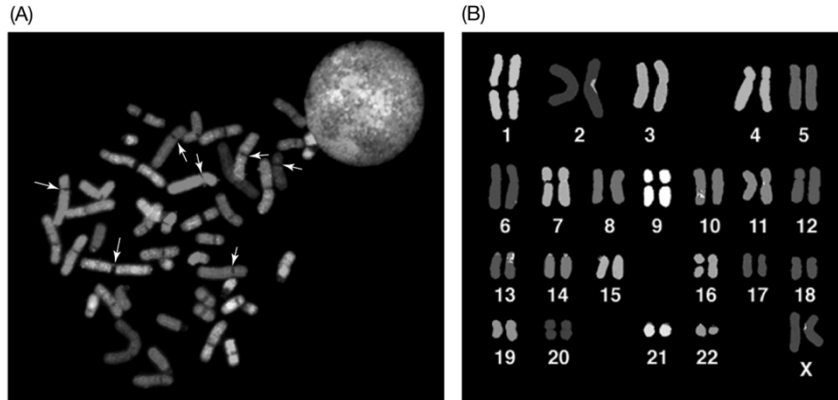
Reproduction: Sexual and Asexual

- In sexually reproducing organisms, certain cells in the adult undergo **MEIOSIS**:
 - ***a diploid cell produces haploid gametes.***
 - Each gamete contains a random mix of one of each pair of homologous chromosomes from the parent.
 - In: **Gonads**
 - animals – testes, ovaries;
 - plant Flowers (anther, ovary)

<http://www.cellsalive.com/meiosis.htm>

Reproduction: Sexual and Asexual

- **Karyotype** = the number, shapes, and sizes of the chromosomes of an organism.



The Human Karyotype

Chromosomes aligned in order, largest to smallest, except Sex chromosomes = always last.

LIFE 9e, Figure 11.16

© 2011 Sinauer Associates, Inc.

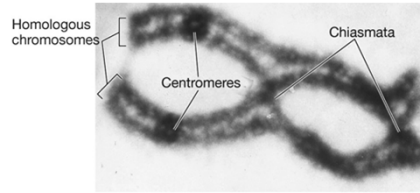
11.7) Meiosis: A Pair of Nuclear Divisions

1. Reduces the chromosome number from diploid to haploid (***Reduction Division!***)
2. Ensures that each haploid cell contains one member of each chromosome pair
 - Preparation for sexual reproduction/fertilization
 - (**$n\text{-mom} + n\text{-dad} \rightarrow 2n$**)
3. Consists of 2 nuclear divisions!

http://highered.mcgraw-hill.com/sites/0072437316/student_view0/chapter12/animations.html# → Stages, etc.

A. Meiosis I

- 1. Prophase I:** homologous chromosomes pair.
 – *material may be exchanged by crossing over between nonsister chromatids of two adjacent homologs*



- 2. Metaphase I:** the paired homologs gather at the equatorial plate.
 – **Each chromosome: has one kinetochore & binds polar microtubules for one pole**

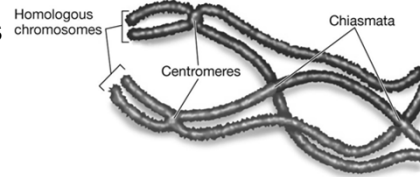
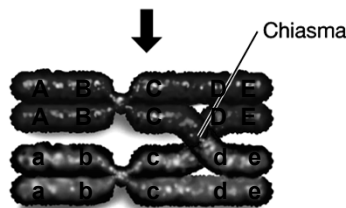
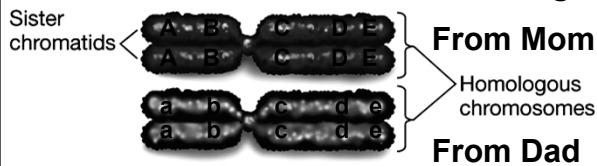


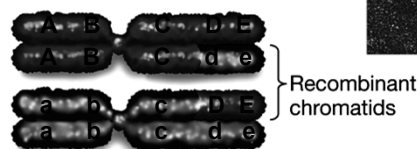
Figure 11.18

- 3. Anaphase I:** entire chromosomes, each with two chromatids, migrate to the poles.
 – ****At end of meiosis I: 2 nuclei, each with the *haploid* number of chromosomes with 2 sister chromatids**

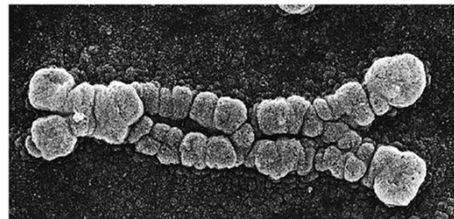
Chiasmata: Evidence of Exchange between Chromatids



Crossing over between **Non-Sister** chromatids



- PROPHASE I:**
 1. Synapsis (pairing)
 2. Crossing over
 3. Chiasmata form

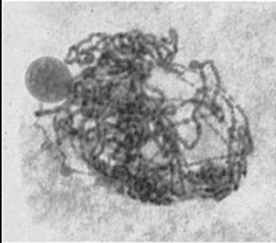


11.19

11.16 Meiosis I (Part 1)

MEIOSIS I

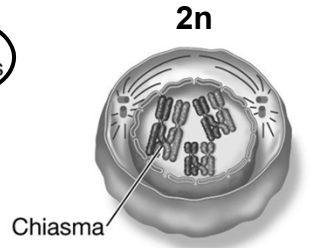
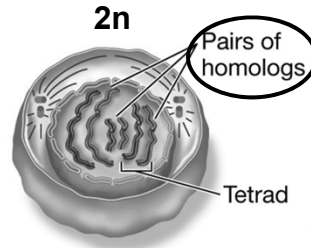
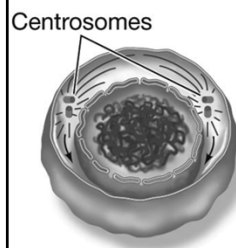
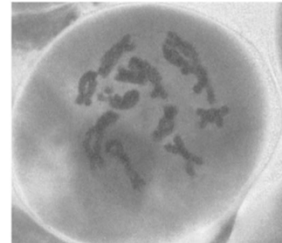
Early prophase I



Mid-prophase I



Late prophase I - Prometaphase



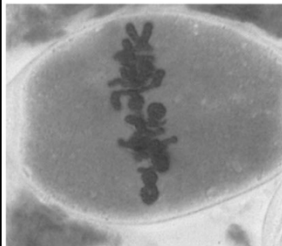
- Synapsis at Chiasmata
- Crossing-over

LIFE 9e, Figure 11.17 (Part 1)

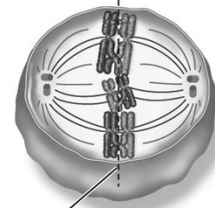
© 2011 Sinauer Associates, Inc.

11.16 Meiosis I (Part 2)

Metaphase I



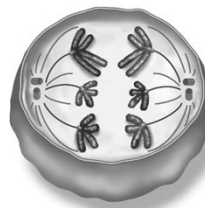
1 kinetochore/chrom.



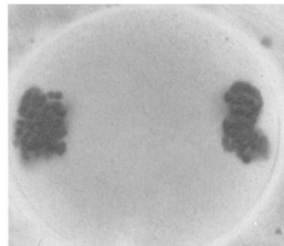
Anaphase I



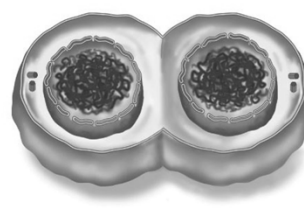
2n



Telophase I



n n



- Recombinant Chromosomes!!

→ **INTERKINESIS** (cell div'n btw meiosis I & II):
• NO DNA Replication!!!!

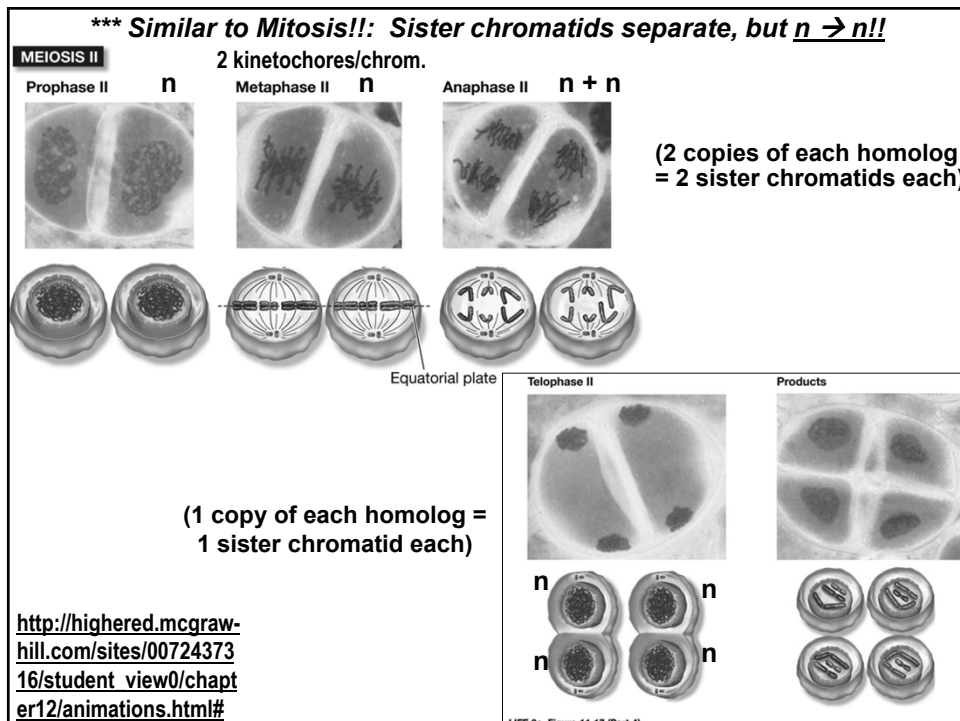
LIFE 9e, Figure 11.17 (Part 2)

B. Meiosis II

1. No DNA replication precedes this division
 - a) (no S phase!)
 - b) *Begins with haploid # of chromosomes*
 - c) *Otherwise similar to mitosis*

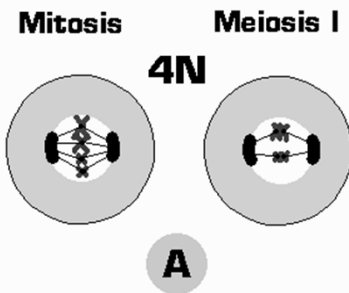
2. Sister chromatids separate

3. **MEIOTIC PRODUCTS = 4 haploid cells!**



C. Mitosis vs. Meiosis

Mitosis is a mechanism for *constancy*:
The parent nucleus produces two daughter nuclei, *identical* to the parent and to each other.



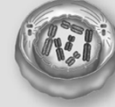
http://highered.mcgraw-hill.com/sites/0072437316/student_view0/chapter12/animations.html#
LIFE 9e, Figure 11.20 (Part 1)

MITOSIS

Parent cell ($2n$)



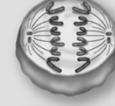
Prophase



Metaphase



Anaphase



Two daughter cells (each $2n$)



MEIOSIS

Parent cell ($2n$)

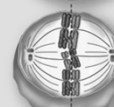


** Prophase I



Pairs of homologs

Metaphase I



** Anaphase I



Telophase I



Interkinesis ... NO DNA Replication!

Four daughter cells (each n)



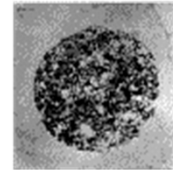
Meiosis is a mechanism for *diversity*:
The parent nucleus produces four haploid daughter nuclei, each *different* from the parent and from its sisters.

LIFE 9e, Figure 11.20 (Part 2)

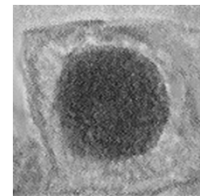
D. Meiosis: Factors Promoting Genetic Diversity

1. Crossing over during Prophase I (*IntRAchrom'l recomb.*)
 - During Synapsis & formation of Chiasmata
 2. Random selection of which homolog of a pair (*from mom or from dad?*) migrates to which pole during Anaphase I (*IntERchromosomal recombination*)
 - → genetic composition of each haploid gamete is different from that of the parent and sisters
 - The more chromosome pairs in a diploid cell, the greater the diversity of chrom. comb'ns generated by meiosis
 - (2^n possibilities! $n = \#$ pairs).
- [**#3.) – & Sex/Fertilization: random combinations of 2 diverse gametes!!!! ... #4.) and choice of partners!?**]

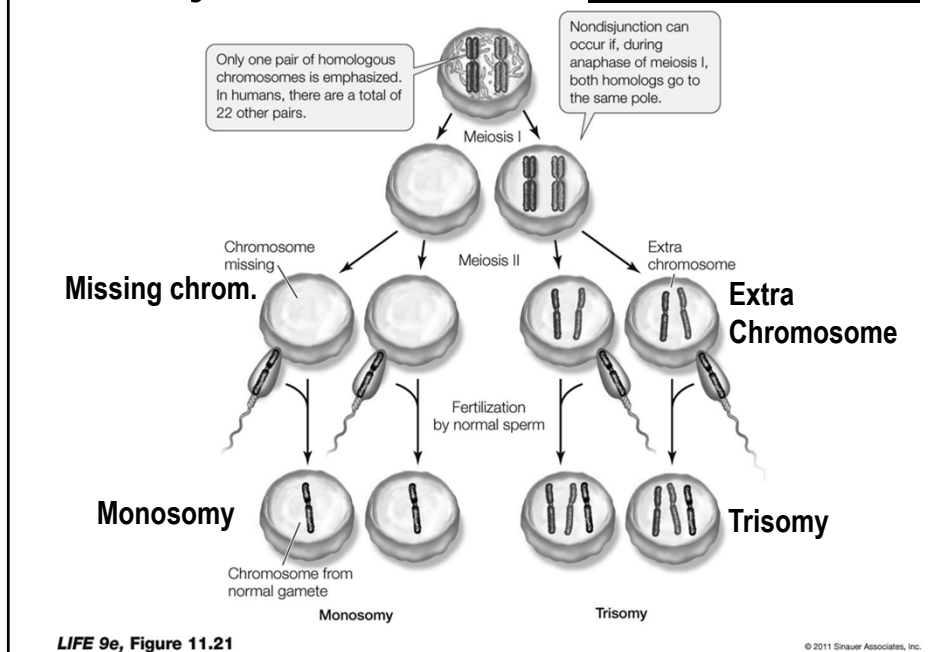
E. Meiotic Errors



- Nondisjunction:
 - one member of a homologous pair fails to separate from the other
 - both go to the same pole
 - → one gamete with an extra chromosome
 - → another other lacking that chromosome
- Fertilization with a normal haploid gamete:
 - results in **aneuploidy** ($2n \pm 1$, etc.)
 - and harmful genetic abnormalities
 - Eg: *Down Syndrome* = Trisomy of Chromosome 21.



Nondisjunction leads to ***ANEUPLOIDY***:



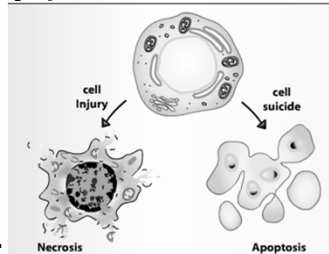
11.8) Cell Death

1. **NECROSIS**: cells damaged by poisons or starved of essential nutrients.

- Swell up and burst.

2. **APOPTOSIS**: a genetically programmed series of events.

- detachment of the cell from its neighbors.
- fragmentation of its nuclear DNA.
 - Development: webbed fingers, nervous system, immune cells
 - Damaged or old cells (risk of DNA mutation, cancer)



<http://www.whfreeman.com/kuby/content/anm/kb04an01.htm>

Apoptosis: Programmed Cell Death

WBC

(A)

A cell in apoptosis displays extensive membrane blebbing.

A normal white blood cell.

(B)

1a External signals can bind to a receptor protein.

1b Internal signals can bind to mitochondria, releasing other signals.

2 Inactive caspase changes its structure to become active.

3 Caspase hydrolyzes nuclear proteins, nucleosomes, etc., resulting in apoptosis.

	NECROSIS	APOPTOSIS
Stimuli	Low O ₂ , toxins, ATP depletion, damage	Specific, genetically programmed physiological signals
ATP required	No	Yes
Cellular pattern	Swelling, organelle disruption, tissue death	Chromatin condensation, membrane blebbing, single-cell death
DNA breakdown	Random fragments	Nucleosome-sized fragments
Plasma membrane	Bursts	Blebbid (see Figure 9.21A)
Fate of dead cells	Ingested by white blood cells	Ingested by neighboring cells
Reaction in tissue	Inflammation	No inflammation

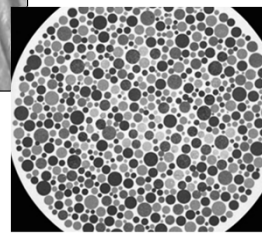
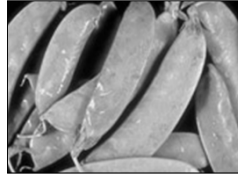
LIFE 9e, Figure 11.22
<http://www.whfreeman.com/kuby/content/anm/kb04an01.htm>
<http://www.whfreeman.com/lodish4e/content/ld01/ld01vs01a.htm>
<http://sites.sinauer.com/cooper6e/animation1702.html>
<http://www.wehi.edu.au/wehi-tv/apoptosis-and-signal-transduction>

Review – Major Themes So Far!!

1. Molecular shape/structure → Molec./Biol. Function
– Lipids, Polysacch., Proteins!..., RNA, DNA
2. In Biological systems: Endergonic processes are COUPLED to Exergonic processes so that they will proceed efficiently.
– ETC/ATP, Active transport, etc.
3. Biological reactions in eukaryotes are compartmentalized.
– glyc, TCA, ETC, lysosome, RER, SER
4. Eukaryotic Gene regulation has MANY levels of **complexity**.
 - *Many steps for each phase of gene expression!*
 - *Each one can be halted in several ways!!*
5. **Mitosis** → generate nuclei identical to each other & original
6. **Meiosis** → generate Haploid nuclei genetically different from each other or from either parent!

Chapter 12: Genetics: Mendel and Beyond

1. The Foundations of Genetics
2. Mendel's Experiments and Laws of Inheritance
3. Alleles and Their Interactions
4. Gene Interactions
5. Genes and Chromosomes
6. Sex Determination and Sex-Linked Inheritance
7. Non-Nuclear (Cytoplasmic) Inheritance



Practice Problems:
<http://fig.cox.miami.edu/~cmallery/150/mendel/problems.htm>

12.1) The Foundations of Genetics

- **Genetics** = the science/study of heredity
- **Ancient Genetic Practices:**
 - “**Artificial Selection**” = Breeding
(humans choose the successful reproducers!!)
 - Dogs bred from Wolves 20,000 years ago!!!.....
 - Ancient Egypt → bred ~400 varieties of dates (palms)
 - Cattle, pets, fruits, vegetables, grains, pets (>5K yrs)





- ❖ **Before Gregor Mendel: “Blended Inheritance”**
 - It was believed that once brought together, the units of inheritance blended and could never be separated.
 - E.G.: **Red**-flowered plant x **Blue**-Flowered Plant → **only Purple Progeny.**
(red and blue traits = “lost”, inseparable once blended; ~melted crayons)
 - **Acquired Characteristics:** gain new traits during life (based on behaviors), and pass them to offspring.
 - **Jean-Baptiste Lamarck**

Controlled Pollination of Plants:

- Facilitated the ease of plant genetics
- Self-crossing & out-crossing possible!!

❖ **Reciprocal Crosses** showed that both parents contributed equally to offspring

- Refuted old theory that one parent contributed more to offspring
- (homunculus in sperm)

RESEARCH METHOD

Anatomy of a pea flower
(shown in long section)

stigma
The stigma is where the pollen lands.

anthers
Anthers at the tip of the stamen are the sites of pollen production.

stamen
Stamens are the male sex organs.

ovary
The ovary is the female sex organ.

Pea flower cross-pollination

Parent plant Parent plant

anthers **stigma**

1 Pollen is transferred from anthers of one flower to the stigma of another flower whose anthers have been stripped off.

2 Seeds are allowed to grow into new plants.

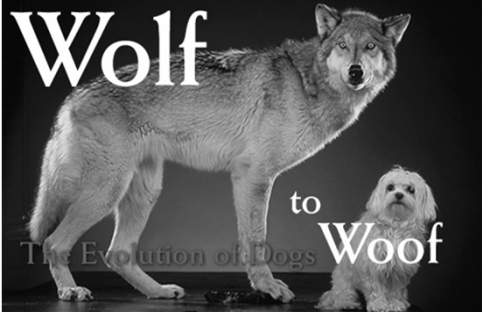
3 Analysis of physical characteristics of the offspring (see Table 10.1) over 2 generations provides evidence of hereditary transmission from both parents.

Pea pod
Seeds (peas)

Fig 10.2

The Foundations of Genetics

- Gregor Mendel's work was **meticulous and well-documented:**
 - 1) Very **quantitative**, with large samples!
 - 2) Used only **DISTINCT traits**, and **TRUE-breeding lines!**
 - 3) Lucky: **no linked traits, no sex-linked, no incomplete dominance**.....
- But his discoveries (1860s) lay dormant until decades later.
 - In 1900, 3 other scientists independently re-proved Mendel's conclusions!!



(National Geographic, 2002)

12.2) Mendel's Experiments & Laws of Inheritance

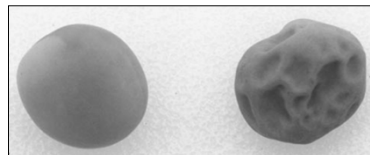
❖ Mendel used garden pea plants for his studies because they were easily cultivated and crossed, and showed numerous characters with clearly DISTINCT traits.

– Used Pure/True-Breeding Strains → showed only one trait for a character after several generations of self-breeding/pollination.

❖ Mendel's SEVEN CHARACTERS in pea plants:

❖ = dominant/recessive trait





























1. **Seed shape** – spherical / wrinkled
2. **Seed color** – yellow / green
3. **Flower color** – purple / white
4. **Pod shape** – inflated / constricted
5. **Pod color** – green / yellow
6. **Flower position** – axial / terminal
7. **Stem height** – tall / dwarf



Mendel's Experiments

- In a Monohybrid Cross, the offspring showed one of the two traits.
- Mendel proposed that the trait observed in the first generation (F₁) was dominant and the other was recessive.

TABLE 12.1

PARENTAL GENERATION PHENOTYPES		F ₂ GENERATION PHENOTYPES		TOTAL	RATIO		
DOMINANT	RECESSIVE	DOMINANT	RECESSIVE				
 Spherical seeds	 Wrinkled seeds			5,474	1,850	7,324	2.96:1
 Yellow seeds	 Green seeds			6,022	2,001	8,023	3.01:1
 Purple flowers	 White flowers			705	224	929	3.15:1
 Inflated pods	 Constricted pods			882	299	1,181	2.95:1
 Green pods	 Yellow pods			428	152	580	2.82:1
 Axial flowers	 Terminal flowers			651	207	858	3.14:1
 Tall stems (1 m)	 Dwarf stems (0.3 m)			787	277	1,064	2.84:1

A. Mendel's Monohybrid Experiments

1) When the F_1 offspring were self-pollinated, F_2 generation showed a **3:1 phenotypic ratio**.

2) the recessive phenotype was present in 1/4 of the offspring.

– *Reappearance of the recessive phenotype refuted the blending hypothesis!!*



HYPOTHESIS When two strains of peas with contrasting traits are bred, their characteristics are irreversibly blended in succeeding generations.

METHOD Plant a true-breeding spherical seed. Plant a true-breeding wrinkled seed.

Parental (P) seeds

1 P plants are cross-pollinated.

Parental (P) plants

Growth

Pollen

F₁ seeds

2 Plant a spherical F₁ seed.

F₁ plant

3 Allow F₁ plants to self-pollinate.

Pollen

RESULTS F₂ seeds from F₁ plant

4 F₂ seeds: 3/4 are spherical, 1/4 are wrinkled (3:1 ratio).

CONCLUSION The hypothesis is rejected. There is no irreversible blending of characteristics, and a recessive trait can reappear in succeeding generations.

Monohybrid Cross:

- Reappearance of **recessive** wrinkled trait in F₂ *disproved blended inheritance!!*
- As did the LACK of an intermediate phenotype in F₁
 - (no “slightly wrinkled” phen)

- **P** = parental generation.
- **F₁** = first filial gen.
- **F₂** = second filial gen.

F₁ seed = spherical = dominant!

F₂ seeds = 1/4 wrinkle, 3/4 spher.

Zygoty

- Because some alleles are dominant and some are recessive, ***the same phenotype can result from different genotypes.***
- 1. **Homozygous genotypes** have two copies of the same allele (AA or aa);
- 2. **Heterozygous genotypes** have two different alleles (Aa).
 - Heterozygous genotypes yield phenotypes showing the dominant trait.
 - AA or Aa show dominant phen.; only aa shows recessive (homozygous recessive)

Mendel's First Law of Inheritance

- ❖ On the basis of many crosses using different characters, Mendel proposed his **First Law, The Law of Segregation:**
 - 1) the units of inheritance (**genes**) are **particulate**,
 - 2) there are two copies/versions (**alleles**) of each gene in every parent, &
 - 3) during gamete formation (meiosis!) the two alleles for a character segregate from each other.
 - **"Particulate Inheritance" – NOT blending!!**

1. The Law of Segregation:

❖ During gamete formation, alleles separate so that each gamete receives only one member of the pair of alleles.

- MONOHYBRID CROSS:

- Traits = separable/particulate

- = distinct!

- Punnett Squares:
consider all possible gametes from each parent.

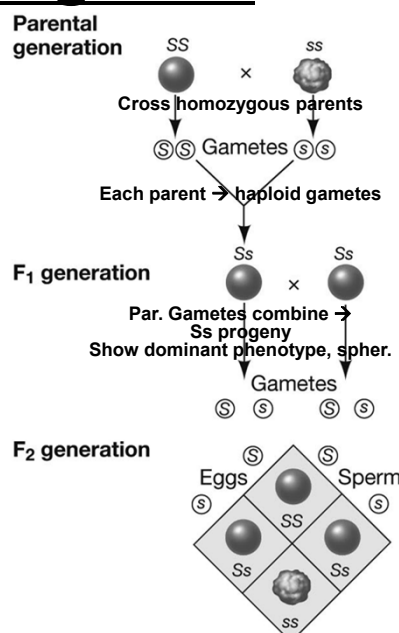


Figure 10/12.4

Modernized Mendelian Genetics: *Meiosis Accounts for Segregation of Alleles!*

- Geneticists who followed Mendel showed that
- 1. Genes are carried on chromosomes, and that
- 2. Alleles are segregated during meiosis I.
 - = modern restatement of Law of Segregation!
 - 1 gene = at 1 locus
 - several alleles may be possible at each locus (for each gene)

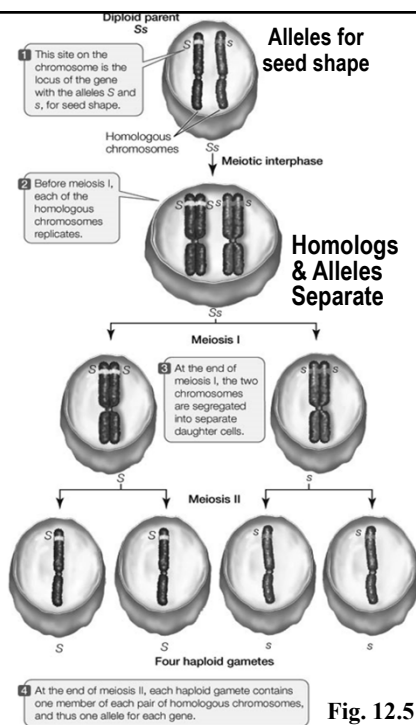
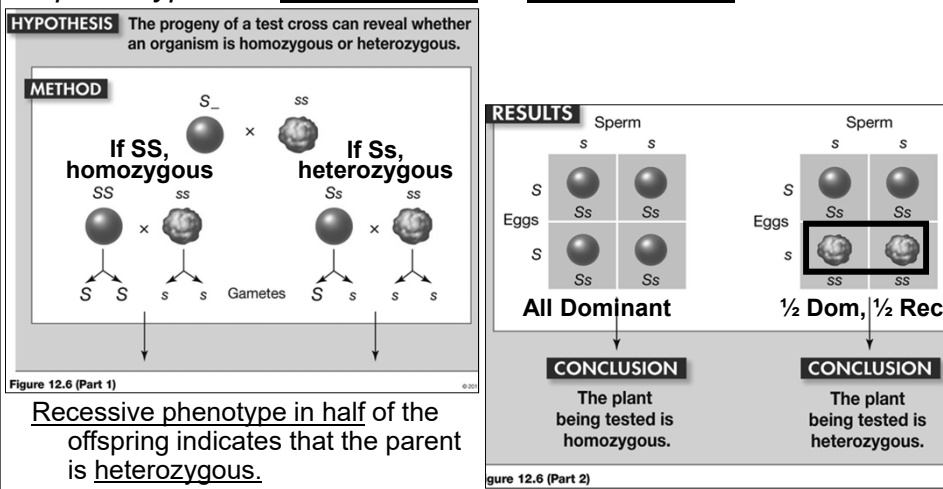


Fig. 12.5

The Test Cross: Homo- or Heterozygous?

- Using a **Test Cross:** (unknown dominant phenotype x homozygous recessive “tester”)

→ determine *whether a plant showing the dominant phenotype was **homozygous** or **heterozygous**.*



B. Mendel's Experiment #2: The DIHYBRID CROSS:

- From studies of the simultaneous inheritance of two characters (genes), Mendel concluded that alleles of different genes assort independently.

- The Law of Independent Assortment**

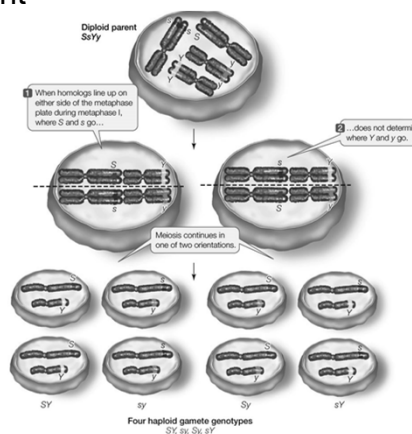


Fig. 12.8: Meiosis Accounts for Independent Assortment of Alleles

12.8

2. Law of Independent Assortment:

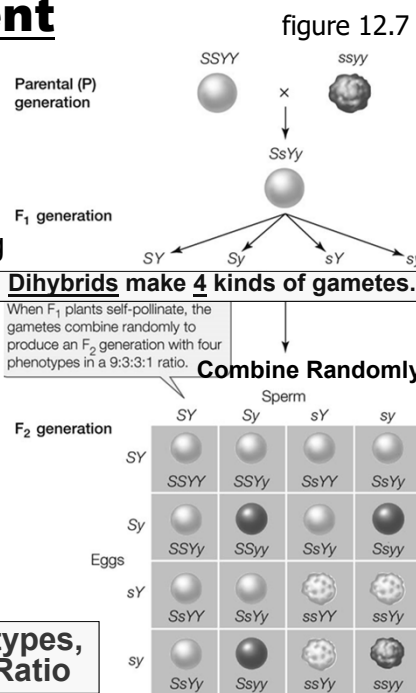
❖ Dihybrid Cross –

- Alleles of different genes assort independently of one another during gamete formation.

- (Not “linked”; Lucky for Mendel!!)
- Genes on different chromosomes!

- **9:3:3:1 = Like two 3:1 ratios superimposed upon each other!!**
 - Each gene’s pair of alleles separated & were distributed independently from the other’s.
 - Each pair of alleles still segregates normally!

**4 Phenotypes,
9:3:3:1 Ratio**



12.3) Genetic Probabilities

- We can predict the results of hybrid crosses by using a Punnett square or by calculating probabilities.

1. **Product Rule:** To determine the joint probability of independent events, individual probabilities are multiplied. (“and”, “also”)
2. **Sum Rule:** To determine the probability of an event that can occur in two or more different ways, they are added. (“or”, “either”)