

9/12/2015

SEPTEMBER 11

INTRODUCTION TO GENETICS BIOL-FRSC 2050H

Fall Semester 2015
Trent University, Peterborough, ON

Cornelya Klütsch
Janet Yee
Debbie Lietz

INTRODUCTION. MITOSIS / MEIOSIS

Contacts

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contact for problems/concerns regarding labs

- Please include the course name/number in the subject line and give your name plus student number in the email.

Biology 2050 H Lab Schedule

Date	Lab Topic	F01	F05
Lab 1 Wednesday Sept 16	9:00 Mitosis & Meiosis	F01	F03
Wednesday Sept 16	13:00	F05	F07
Wednesday Sept 16	17:00		F09
Lab 1 Wednesday Sept. 23	9:00 Mitosis & Meiosis	F02	F04
Wednesday Sept. 23	13:00	F06	F08
Wednesday Sept. 23	17:00		F10
Lab 2 Wednesday Sept. 30	9:00 Mendelian Genetics	F01	F03
Wednesday Sept. 30	13:00	F05	F07
Wednesday Sept. 30	17:00		F09
Lab 2 Wednesday Oct. 07	9:00 Mendelian Genetics	F02	F04
Wednesday Oct. 07	13:00	F06	F08
Wednesday Oct. 07	17:00		F10

- You can only attend the lab you have signed up for

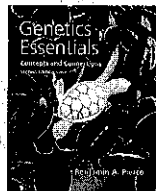
- Assignments must be handed in at the end of each lab

F02

Required textbook

- Genetic Essentials: Concepts & Connections 2nd edition (2013) by Benjamin Pierce, WH Freeman, New York.

- 1st edition of this textbook allowed
- Textbook will be used in lab sections
- Worked problems and concept checks



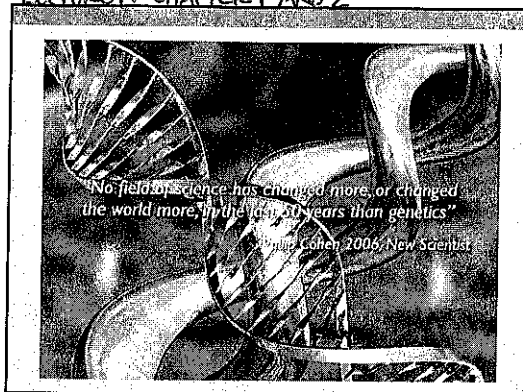
Evaluation

Type of Assignment	Weighting	Due Date
Quiz 1	11.5%	Fri., Oct. 02
Test 1 (all material in Dr. Klütsch's section)	26%	Fri., Oct. 23
Quiz 2	10%	Fri., Nov. 20
Test 2* (all material in Dr. Yee's section)	26%	Dec. 5 - 20*
iClicker	1.5%	Dr. Yee's section only
Labs (5 labs each worth 5%)*	25%	See Lab Schedule
Total	100%	

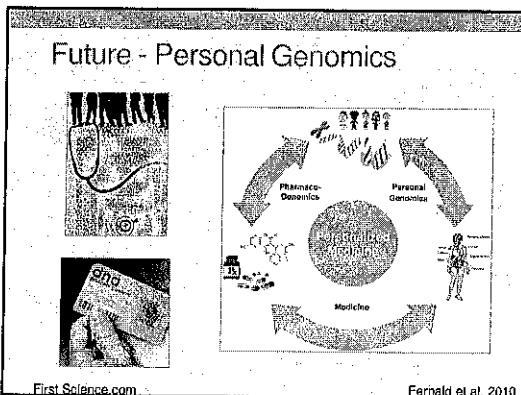
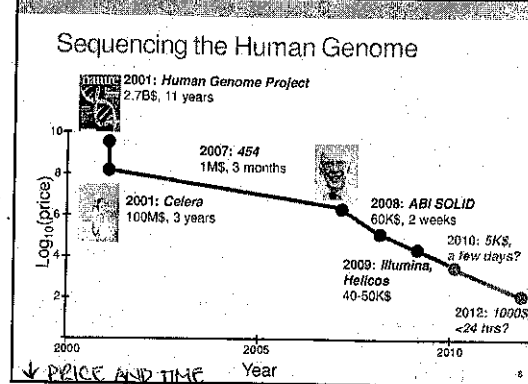
- There are no makeup quizzes/tests scheduled!

- Quizzes/tests are not transferable – you can't get higher % in one of Dr. Yee's tests to make up for missed quizzes/tests

LECTURE 1. CHAPTER 1 AND 2



GENOME-COMPLETE SET OF GENETIC INSTRUCTIONS



Agriculture

Replacing pesticides with genetic engineering

Diamondback moths

Disease-resistant crops/vegetables

Innate™ Conventional
Disease-resistant potato

GENE THERAPY - THE DIRECT ALTERATION OF GENES TO TREAT HUMAN DISEASES.

Forensics & Wildlife

AdaptTree
Assesses the adaptive strength of populations across geographic time

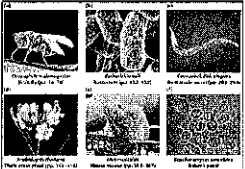
Socio-economic and ethical implications

- More food for a growing human population
- Diseases are better treatable
- Privacy issues (e.g., should an employer get your information?)
- Health insurance
- Long-term effects: what are long-term effects of altering the genome of individuals/populations?

ALL ORGANISMS ARE SIMILAR GENETIC SYSTEMS, AND GENETIC VARIATION IS THE FOUNDATION OF THE DIVERSITY OF ALL LIFE.

1. ALL LIFE FORMS ARE GENETICALLY RELATED
2. RESEARCH FINDINGS ON ONE'S ORGANISM GENE FUNCTION CAN OFTEN BE APPLIED TO OTHER ORGANISMS.
3. GENES FROM ONE ORGANISM CAN OFTEN EXIST AND THRIVE IN ANOTHER ORGANISM.

Model organisms

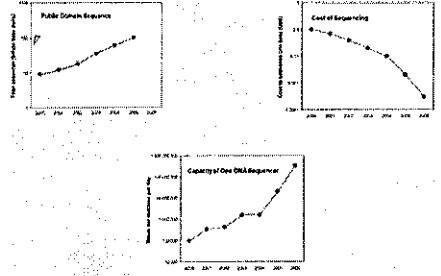


- Short generation time
- Manageable number of offspring
- Lab environment
- Low cost
- (small genome and/or large databases)

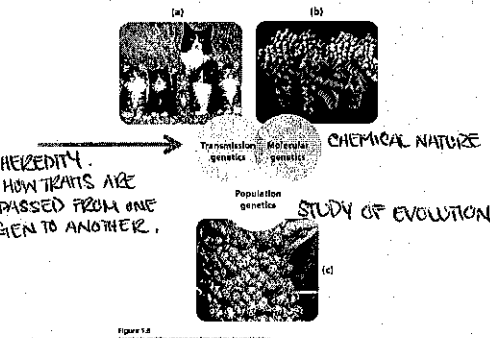
FRUIT FLY, E. COLI, NEMATODE (ROUNDWORM), THALE-CRESS PLANT, MOUSE, BAKER'S YEAST.

CHAPTER 1 INTRODUCTION OF GENETICS

Next Generation Sequencing



DIVISIONS OF GENETICS

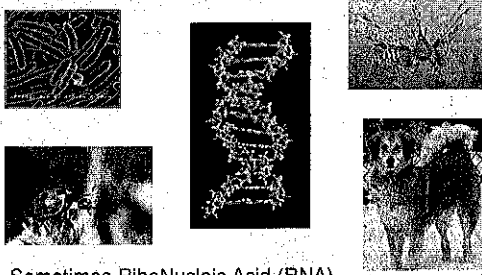


HEREDITY - HOW TRAITS ARE PASSED FROM ONE GEN TO ANOTHER.

CHEMICAL NATURE

STUDY OF EVOLUTION

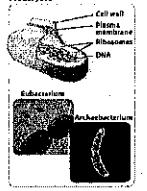
DeoxyriboNucleic Acid (DNA)



Sometimes RiBoNucleic Acid (RNA)

DNA
↓
(TRANSCRIPTION)
↓
RNA
(TRANSLATION)
↓
PROTEIN


PROKARYOTIC CELLS



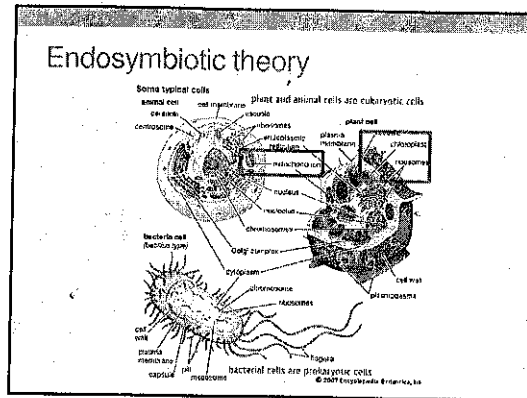
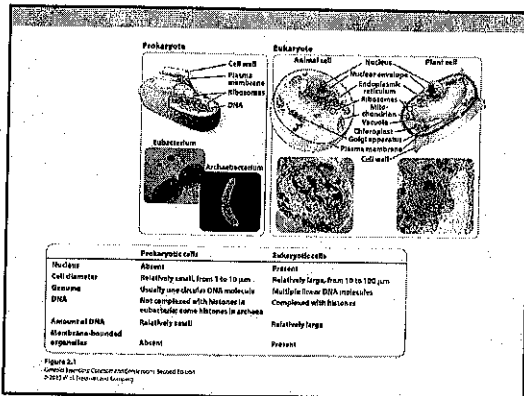
- Unicellular, with no compartmentalized cell structure
- Prokaryotic DNA does not exist in the highly ordered and packed arrangement
- Made up of eubacteria and archaea

Prokaryotic cells	
Nucleus	Absent
Cell diameter	Relatively small, from 1 to 5 µm
Genome	Usually one closed DNA molecule
DNA	Not associated with histones to produce the same chromatin structure
Amount of DNA	Relatively small
Membrane-bound organelles	Absent

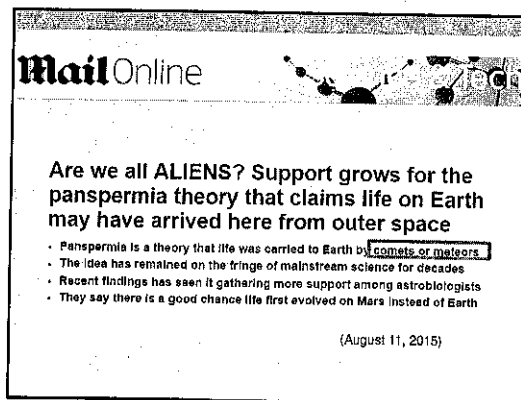
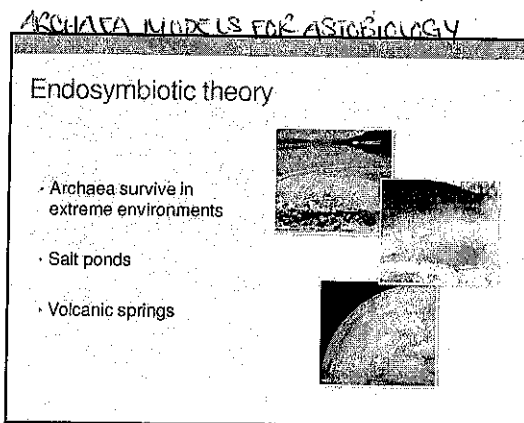
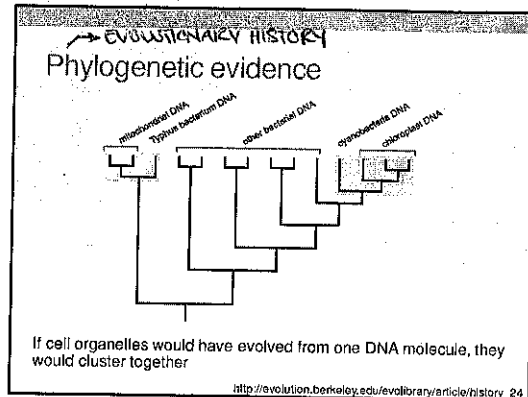
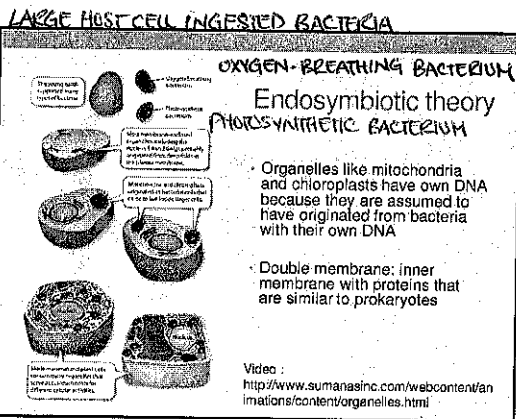
NO NUCLEUS.



NO MEMBRANE-BOUND ORGANELLES.
ONE CIRCULAR DNA MOLECULE.



EVOLUTIONARY THEORY THAT EXPLAINS THE ORIGIN OF EUKARYOTIC CELLS FROM PROKARYOTES



GENOTYPE GENES THAT DETERMINE TRAIT
PHENOTYPE TRAIT THAT THEY PRODUCE.

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CHAPTER 2 CHROMOSOMES AND CELLULAR REPRODUCTION

Eukaryotes: Chromosomes MADE UP OF NUCLEIC ACIDS AND PROTEINS

Chromosomes are the central packaged units by which DNA is transmitted to new cells or generations.

GENES ARE LOCATED IN THE CHROMOSOMES.

2.1. PROKARYOTIC AND EUKARYOTIC CELLS DIFFER IN A NUMBER OF GENETIC CHARACTERISTICS

LINEAR DNA

Figure 2.3a
Quail, L. S. and C. M. (1978) *Genetics and Evolution*, 2nd Edition, © 1978, W. H. Freeman and Company

NUMBER AND APPEARANCE OF CHROMOSOMES IN THE NUCLEUS OF A EUKARYOTIC CELL

Karyotype: complete set of chromosomes

Humans have 23 pairs of chromosomes.

A diploid organism has two sets of chromosomes organized as homologous pairs.

Allele A
Allele a

These two versions of a gene encode a trait such as hair color.

Figure 2.8
Copyright 2011 Sinauer Associates, Inc. and W. H. Freeman & Co.

Chromosome number in different species

Common Name	Species	Diploid number	Common Name	Species	Diploid number
Humans	<i>Homo sapiens</i>	46	Corn	<i>Zea mays</i>	20
Monkeys	<i>Rhesus macaca</i>	42	Potato	<i>S. tuberosum</i>	48
Dog	<i>Canis familiaris</i>	78	Green algae	<i>A. muscovata</i>	20
Cat	<i>Felis domesticus</i>	38			
Mouse	<i>Mus musculus</i>	40	Fungus (2n)		
Frog	<i>Rana pipiens</i>	26	Yeast	<i>S. cerevisiae</i>	32
Fruit fly	<i>Drosophila melanogaster</i>	8	Fungus (1n)		Haploid number
Flaxweed	<i>Phlox pilularis</i>	18	Mold	<i>Penicillium species</i>	4

HOMOLOGOUS PAIR 2 CHROMOSOMES USUALLY ANKE IN STRUCTURE AND SIZE. EACH CARRIES GENETIC INFO FOR THE SAME SET OF HEREDITARY CHARACTERISTICS.

Autosomes and sex chromosomes

Autosomes: Paired chromosomes with the same length/shape
Example: body size, hair colour

Sex chromosomes (ALLOSOME): Chromosome pair that differs in size/shape
Sex determination

XX FEMALE XY MALE

Chromosomes

All times a chromosome consists of a single chromatid...
...at other times, it consists of two (sister) chromatids.

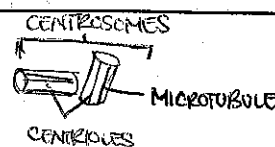
The telomeres are the stable ends of chromosomes.

Kinetochore: PROTEIN
Spindle microtubules

The centromere is a constricted region of the chromosome where the kinetochores form and the spindle microtubules attach.

Figure 2.6
Copyright 2011 Sinauer Associates, Inc. and W. H. Freeman & Co.

FUNCTIONAL CHROMOSOMES
• CENTROMERES, TELOMERES, AND ORIGINS OF REPLICATION



FOR ANY CELL TO REPRODUCE SUCCESSFULLY

1. GENETIC INFO. MUST BE COPIED
2. COPIES OF GENETIC INFO. MUST BE SEPARATED FROM EACH OTHER
3. THE CELL MUST DIVIDE

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Chromosomes types

- Metacentric: centromere in the middle
- Submetacentric: between middle and end
- Acrocentric: close to the end
- Telocentric: at the end

How is DNA transmitted?

- Mitosis: cell division – production of two cells that are identical to the parent cell
- Meiosis: production of 4 cells with 1/2 the number of chromosomes as the parent cell that are genetically different

Mitosis (chapter 2)

PROKARYOTIC CELL REPRODUCTION

- CIRCULAR CHROMOSOME REPLICATED
- ORIGIN OF REPLICATION

MITOSIS - EUKARYOTIC CELL REPRODUCTION

REQUIRES MULTIPLE DNA MOLECULES

Mitosis – cell cycle

During G₁, the cell grows.

In S, DNA replicates.

In G₂, the cell prepares for mitosis.

In phase, nuclear and cell division.

During M, cytokinesis occurs.

Cells may enter G₀, a non-dividing phase.

After the G₁/S checkpoint, the cell is committed to dividing.

After the G₂/M checkpoint, the cell is committed to dividing.

Spindle assembly checkpoint.

Mitosis and cytokinesis (cell division) take place in M phase.

Cancer UNCONTROLLED GROWTH.

- ### METAPHASE CHECKPOINT
- PASS THIS CP IF:
- ALL CHROMOSOMES ARE ATTACHED TO SPINDLE APPARATUS.
 - G₁ CHECKPOINT
 - CELL SIZE IS ADEQUATE
 - NUTRIENTS ARE SUFFICIENT
 - SOCIAL SIGNALS ARE PRESENT
 - DNA IS UNDAMAGED
 - G₂ CHECKPOINT
 - CHROMOSOMES HAVE REPLICATED SUCCESSFULLY
 - DNA IS UNDAMAGED
 - ACTIVATED MPF IS PRESENT.

FAIL CHECKPOINT - APOPTOSIS

Cancer

METAPHASE CHECKPOINT

- Early cancer treatment: Colchicine - INHIBITS MICROTUBULE POLYMERIZATION - Interrupts cell cycle
- Cells are unable to divide
- Toxic side effects

THE G₁ CELL-CYCLE CHECKPOINT

FIGURE 11.12 The Three Cell-Cycle Checkpoints. Source: Mastering Biology (Pearson 2011)

2-2 CELL REPRODUCTION REQUIRES THE COPYING OF THE GENETIC MATERIAL, SEPARATION OF THE COPIES, AND CELL DIVISION.

Interphase: Chromosomes relaxed, visible only as diffuse chromatin. Nuclear envelope is present and chromosomes are relaxed.

Prophase: Chromosomes condense. Each chromosome possesses two chromatids. The mitotic spindle forms.

Prometaphase: The nuclear membrane disintegrates. Spindle microtubules attach to chromatids.

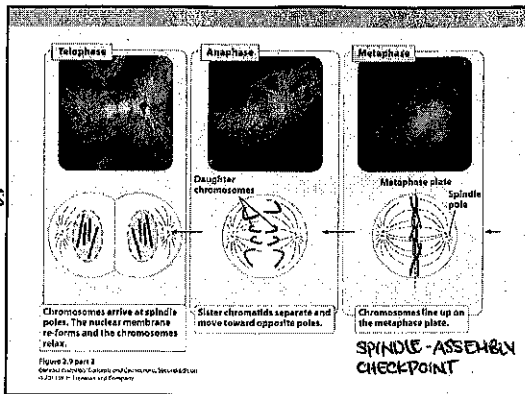
- G₀ PHASE STABLE, NONDIVIDING PERIOD
- ### INTERPHASE
- G₁ GROWTH / DEVELOPMENT, G₁ CP
 - S SYNTHESIS OF DNA
 - G₂ PREPARATION FOR DIVISION, G₂ CP

- PROPHASE BECOME VISIBLE UNDER A LIGHT MICROSCOPE
- CHROMOSOMES CONDENSE, MITOTIC SPINDLE FORMS
- PROMETAPHASE NUCLEAR ENVELOPE DISINTEGRATES
- SPINDLE MICROTUBULES ANCHOR TO KINETOCHORES

NATURATION PROMOTING FACTOR.

CYTOKINESIS

- CYTOPLASM DIVIDES
- CELL WALLS FORMS IN PLANT



10 MINUTE BREAK

2.3 SEXUAL REPRODUCTION PRODUCES GENETIC VARIATION THROUGH THE PROCESS OF MEIOSIS

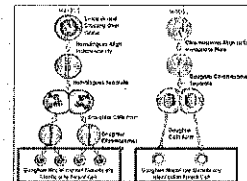
MEIOSIS

SEXUAL REPRODUCTION

1. MEIOSIS
2. FERTILIZATION

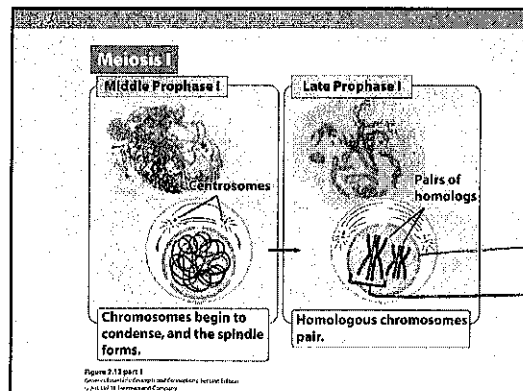
How is DNA transmitted?

- Mitosis: cell division – production of two cells that are identical to the parent cell
- Meiosis: production of 4 cells with 1/2 the number of chromosomes as the parent cell that are genetically different

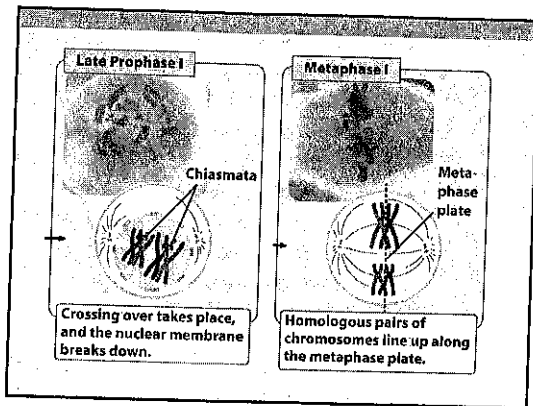


- INTERPHASE 2N (1)
- PROPHASE I 4N (2)
- METAPHASE I 4N (X)
- ANAPHASE I 4N (X)
- TELOPHASE I CYTOKINESIS (X)
- PROPHASE II 2N (3)
- METAPHASE II 2N (X)
- ANAPHASE II 2N (X)
- TELOPHASE II CYTOKINESIS N (4)

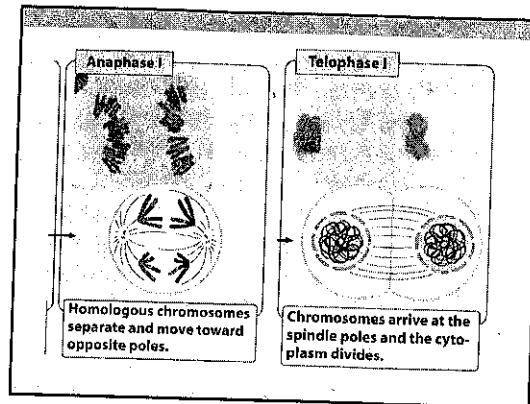
Meiosis (chapter 2)



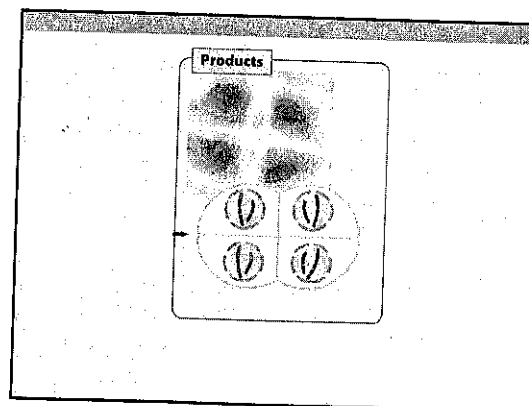
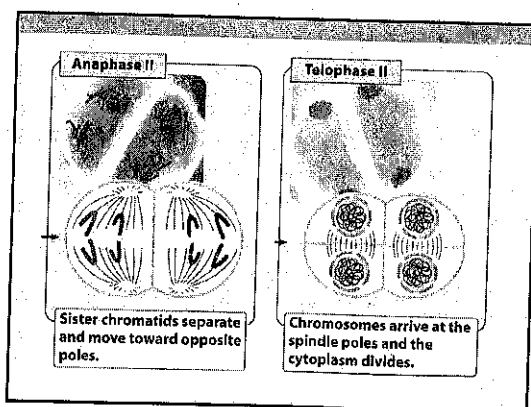
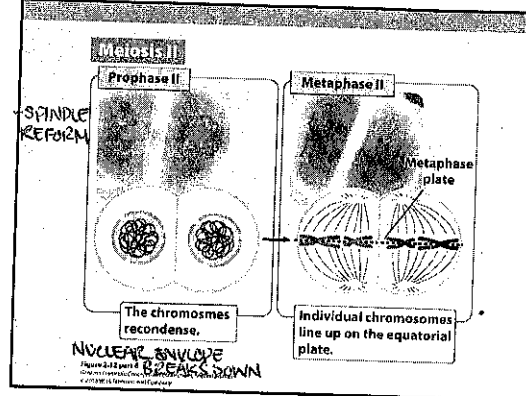
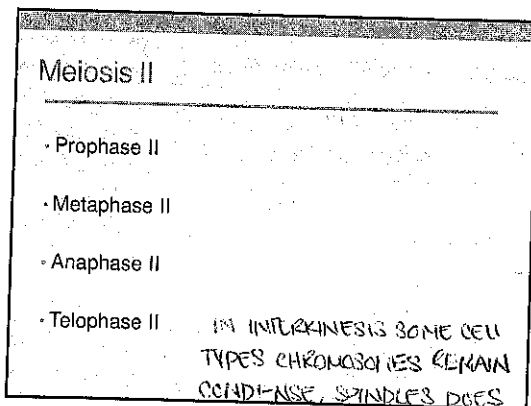
PAIR UP AND BEGIN SYNAPSIS. VERY CLOSE ASSOCIATION 4 CHROMATIDS (BIVALENT OR TETRAD)



CROSSING OVER HOMOLOGOUS CHROMOSOMES EXCHANGE GENETIC INFORMATION



INTERKINESIS PERIOD BETWEEN MEIOSIS I AND II - NUCLEAR MEMBRANE RE-FORMS AROUND THE CHROMOSOMES, SPINDLES BREAK DOWN, AND CHROMOSOMES RELAX



SOURCES OF GENETIC VARIATION IN MEIOSIS

Genetic variation

- Individual chromosomes line up in mitosis, whereas homologous chromosome pairs line up in meiosis

↓

Genetic variation

- Crossing over in late prophase I
- Independent assortment during metaphase I

Recombination – crossing over

1 One chromosome possesses the A and B alleles...
 2 ...and the homologous chromosome possesses the a and b alleles.
 3 DNA replication in the S phase produces identical sister chromatids.
 4 During crossing over in prophase I, segments of non-sister chromatids are exchanged.
 5 After meiosis I and II, each of the resulting cells carries a unique combination of alleles.

Figure 2.13
 Campbell, Reece, and Mitchell, *Biological Science*, 6th Edition
 © 2011 Sinauer Associates, Inc.

Random assortment

1 The first pair of homologous chromosomes separates.
 2 The second pair of homologous chromosomes separates.
 3 The third pair of homologous chromosomes separates.

Figure 2.14
 Campbell, Reece, and Mitchell, *Biological Science*, 6th Edition
 © 2011 Sinauer Associates, Inc.

THERE ARE 4 POSSIBLE WAYS FOR 3 PAIRS TO ALIGN.

- Crossing over shuffles alleles on the same chromosome into new combinations
- Independent assortment shuffles alleles on different chromosomes

MEIOSIS IN ANIMALS

SPERMATOGENESIS (Male gametogenesis)

OOGENESIS (Female gametogenesis)

Figure 2.17
 Campbell, Reece, and Mitchell, *Biological Science*, 6th Edition
 © 2011 Sinauer Associates, Inc.

Table 2.3 Mitosis, meiosis I, and meiosis II compared

Event	Mitosis	Meiosis I	Meiosis II
Cell division	Yes	Yes	Yes
Chromosomal reduction	No	Yes	No
Genetic variation produced	No	Yes	No
Crossing over	No	Yes	No
Random distribution of maternal and paternal chromosomes	No	Yes	No
Metaphase	Individual chromosomes line up	Homologous pairs line up	Individual chromosomes line up
Anaphase	Chromatids separate	Homologous chromosomes separate	Chromatids separate

Table 2.3
 Campbell, Reece, and Mitchell, *Biological Science*, 6th Edition
 © 2011 Sinauer Associates, Inc.

- SPERMATOGONIUM (2n)
 ↓ PROPHASE I, BECOME SPERMATOCYTE
 PRIMARY SPERMATOCYTE (2n)
 ↓ MEIOSIS I, 2NDARY SPERMATOCYTE
 SECONDARY SPERMATOCYTE (2n)
 ↓ MEIOSIS II, PRODUCE 4 HAPLOID
 SPERMATID (1n)
 MATURATION

- OOGONIUM (2n)
 ↓ PROPHASE I
 PRIMARY OOCYTE (2n)
 ↓ MEIOSIS I, 1ST POLAR BODY (DISINTEGRATES)
 SECONDARY OOCYTE (n)
 ↓ MEIOSIS II < OVUM
 OVUM (1n)

Errors in mitosis

Nondisjunction in mitosis

- Nondisjunction
- Mosaic genomes *
- Fatal if early in development

* DENOTES THE PRESENCE OF 2 OR MORE POPULATIONS OF CELLS W/ DIFF GENOTYPES IN ONE INDIVIDUAL

Figure 6.16a
Developmental Biology of Vertebrates, Second Edition
© 2011 W. H. Freeman and Company

NONDISJUNCTION THE FAILURE OF HOMOLOGOUS CHROMOSOMES OR SISTER CHROMATIDS TO SEPARATE DURING CELL DIVISION

Nondisjunction in meiosis I

MEIOSIS I → **Gametes** → **MEIOSIS II** → **Fertilization** → **Zygotes**

Normal gamete → Normal diploid (2n)

Nondisjunction → Trisomic (2n + 1) / Monosomic (2n - 1)

Figure 6.16a
Developmental Biology of Vertebrates, Second Edition
© 2011 W. H. Freeman and Company

Nondisjunction in meiosis II

MEIOSIS I → **Gametes** → **MEIOSIS II** → **Fertilization** → **Zygotes**

Normal gamete → Normal diploid (2n)

Nondisjunction → Trisomic (2n + 1) / Monosomic (2n - 1)

Figure 6.16b
Developmental Biology of Vertebrates, Second Edition
© 2011 W. H. Freeman and Company

Types of Aneuploidy

PRESENCE OF AN ABNORMAL NUMBER OF CHROMOSOME

- **Nullisomy:** loss of both members of a homologous pair of chromosomes. $2n - 2$
HUMANS WILL NOT SURVIVE **NONDISJUNCTION**
- **Monosomy:** loss of a single chromosome. $2n - 1$
 $n - 1 + n \rightarrow 2n - 1$
- **Trisomy:** gain of a single chromosome. $2n + 1$
 $n + 1 + n \rightarrow 2n + 1$
- **Tetrasomy:** gain of two homologous chromosomes. $2n + 2$ **48, XXXX SYNDROME**

Multiple choice example questions

Which is the correct order of stages in the cell cycle?

- G1, S, prophase, metaphase, anaphase
- S, G1, prophase, metaphase, anaphase
- prophase, S, G1, metaphase, anaphase
- S, G1, anaphase, prophase, metaphase

Note that the term mitosis is not given here but the term cell cycle clearly identifies this question to refer to mitosis and not to meiosis.

Multiple choice example questions

How many chromosomes would have been in an oogonium that produced an ovum with 18 chromosomes?

- 2
- 9
- 18
- 36
- 72

ovum is haploid
oogonium is diploid
2x