Biology 30 IB
Cells, Chromosomes, & DNA

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Resources

- Videos and Animations

Contents

Terms

- **Somatic cells** are all cells in the body except sex cells — sperm and egg cells
- A human somatic cell has 46 chromosomes
- **Cell division** is done by Eukaryotic cells — have a nucleus
- **Binary fission** is done by Prokaryotic cells — have no nucleus, such as bacteria

(17.1) Cell Division

Purpose

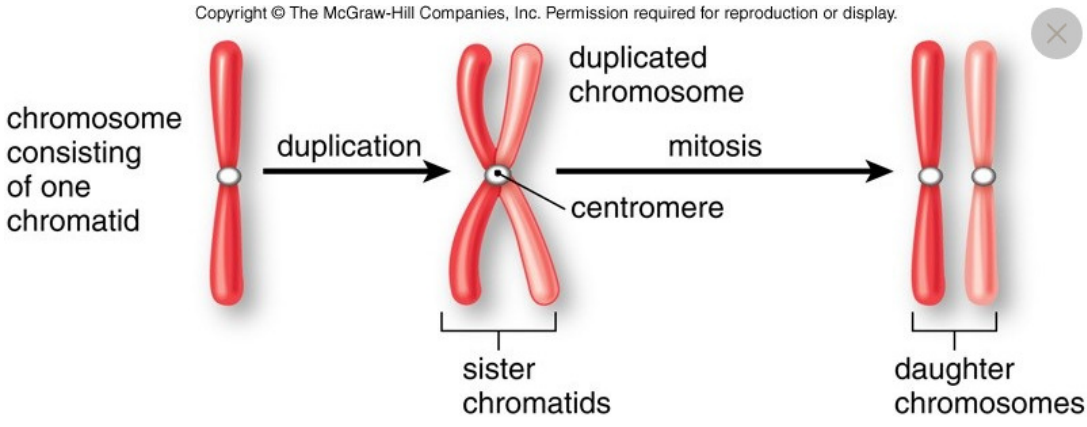
- Unicellular organisms (i.e. zygote) $\longrightarrow$ Multicellular organisms
- Growth and maintenance of body cells — replacement of worn out cells

Chromosomes

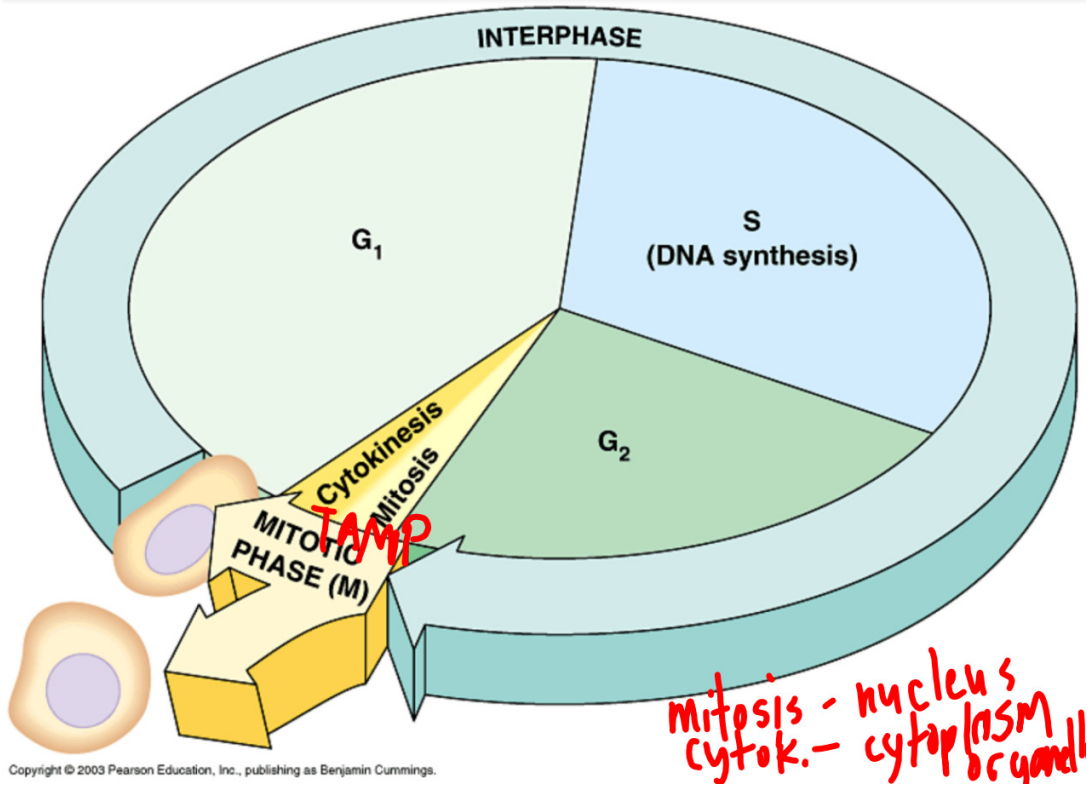
- Comprised of...
  - nucleic acids (DNA)
  - proteins
- Either...
  - **Uncondensed** aka. **Chromatin** = long, thin strands. invisible to microscope
  - **Condensed** = thick & shortened. visible to microscope

Chromatid

- The strand that makes up a normal chromosome
- In mitosis...
  - A chromosome duplicates into two identical chromatids, joined together by a **centromere**, to form a **duplicated chromosome**
  - These chromatids are referred as **sister chromatids** in this state
  - Each chromatid of a duplicated chromosome goes to each of the two new cells



Cell Cycle



A continuous cycle that involves all steps of a cell's life, especially cell division.

Interphase

MAJOR PHASE

- 90% of cell cycle
- All cell activity when not dividing

Gap 1 (G_1)

- Cell growth and general function
- After cell division, cells may be smaller than their parent. Cell growth is needed

S Phase (S)

- DNA is doubled
- Single(-chromatid) chromosome $\xrightarrow{\text{duplication}}$ double(-chromatid) chromosome

Gap 2 (G_2)

- Organelles are doubled, and proteins for the new cell are produced

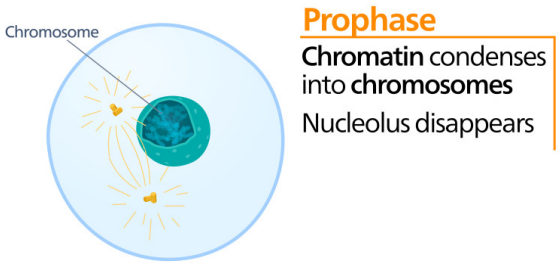
Mitotic Phase

MAJOR PHASE; occurs in somatic cells.

Distribution of nucleus and its contents.

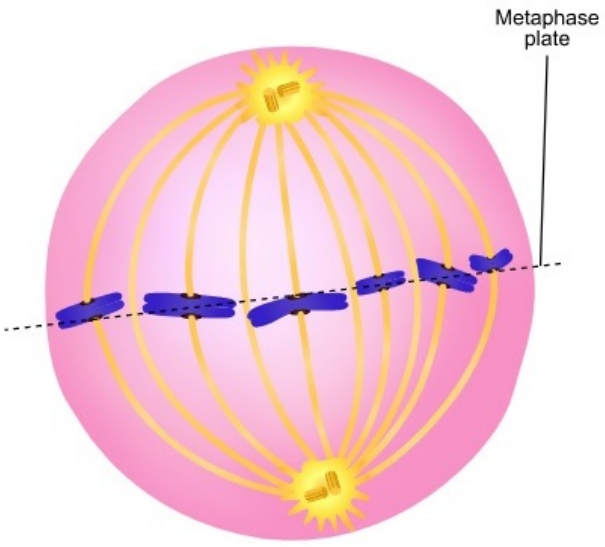
Prophase

- Chromatin condense — shorten & thicken — into chromosomes, becoming visible
- Nuclear membrane fades
- Animal cells only...
  - **Centrioles** (aka. centrosomes) move to opposite poles of cell. (N/S, E/W)
  - Two centrioles are at each pole, total four, for each cell
  - Centrioles deploy **spindle fibers**
- Without centrioles — such as plant cells — spindle fibers are still present and the cycle works the same



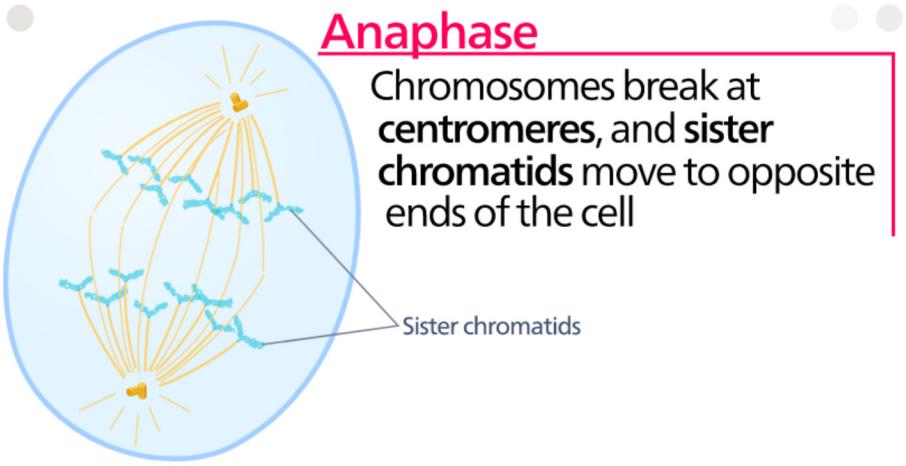
Metaphase

- **Equatorial plate** = center of cell
- Sister chromatids move towards equatorial plate
- Chromosomes attach to spindle fibers



Anaphase

- Centromeres divide
- (Now) chromatids move towards spindle fibers — i.e. opposite poles of cell



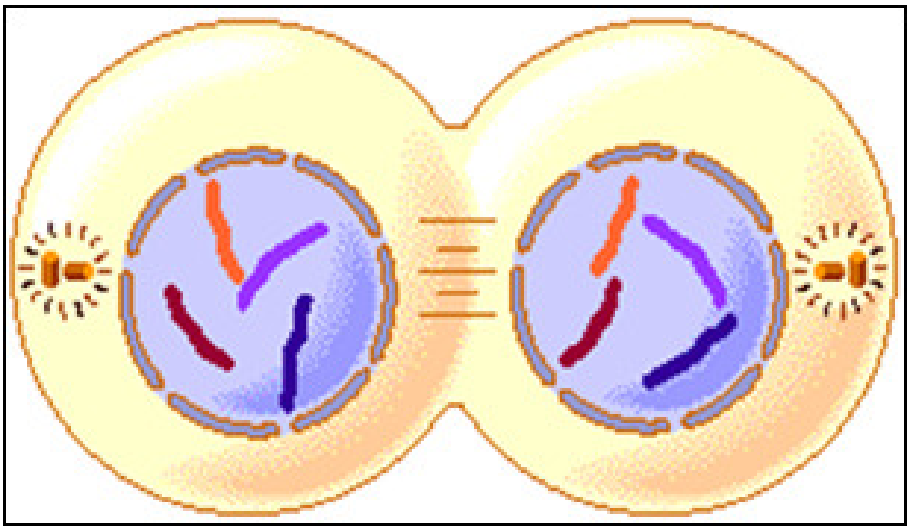
Telophase

- Spindle fibers dissolve
- Nuclear membrane forms around each mass of chromatin

Cytokinesis

Technically occurs at the end of telophase.

- Division of cytoplasm and distribution of organelles to "daughter" cells
- Involves **cleavage**, pinching off in the center as the cytoplasm moves to opposite poles
- In plant cells only, a **cell plate** is distributed, which develops into a new cell wall



Cell Properties

Biological Clock

Immature cells always have 50 division, regardless of...

- duration frozen
- stage/phase that cell division was suspended

Death & Aging

Cells may stop dividing due to...

- **Senescence** = aging, irreversible changes that eventually lead to death
- **Specialization** = the more specialized/differentiated a cell is, the less likely it will undergo mitosis

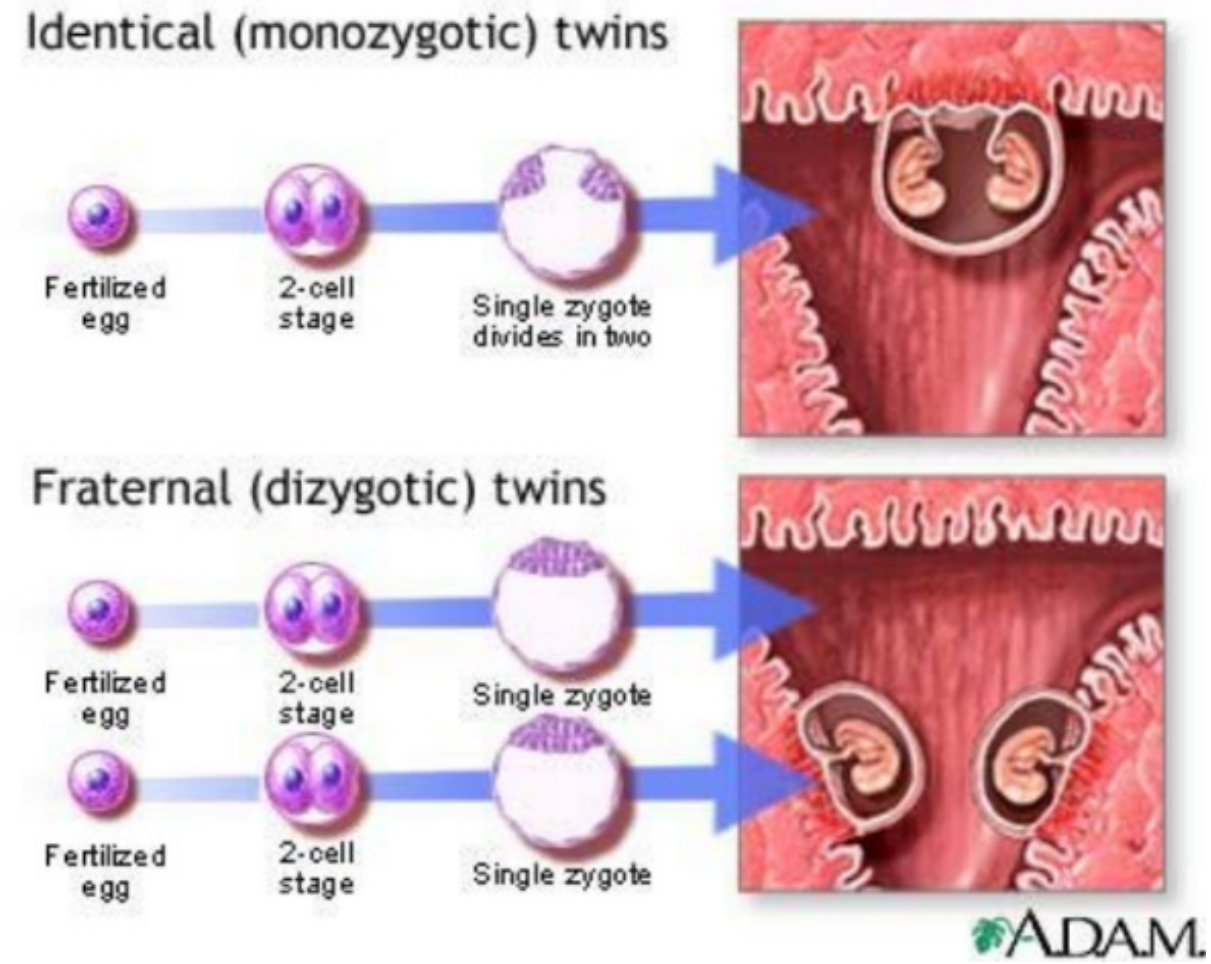
Cells that avoid aging are...

- **Spermatogonia** = sperm-producing cells, immature & unspecialized
- Cancer cells of a tumor, which do not become specialized

(17.2) Natural Cloning

- Asexual/nonsexual reproduction
- Identical offspring from a single cell

Twins



Identical Twins

- Originate from single egg cell
- During mitosis, one of the cells breaks free; this cell forms a 2nd embryo
- If cell clusters remain separate, two babies with identical gene structures will develop
- Same gender, blood type, similar facial structure (nature vs. nurture)

Fraternal Twins

- Two different eggs fertilized by different sperm cells
- Not to be confused with identical twins — do not have identical genes

Unnatural Cloning

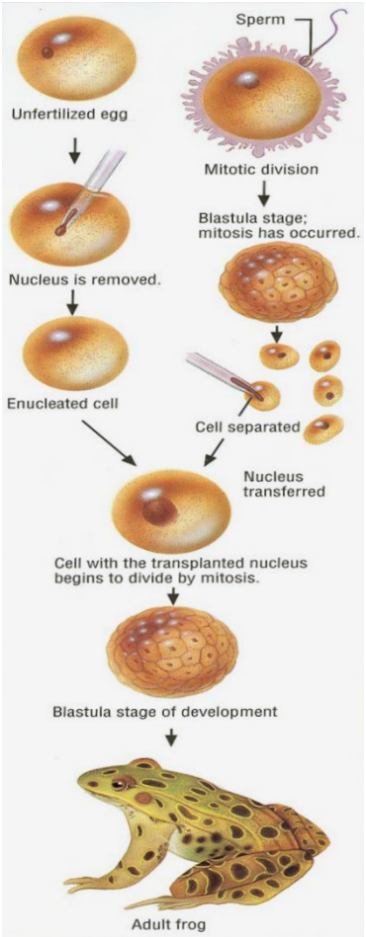
A **totipotent** nucleus is a nucleus that is able to bring a cell from egg to adult.

Plant Cloning

- useful, since cloned plants have predictable characteristics
- requires delaying cell specialization

Animal Cloning

- With a micropipette, the nucleus is extracted from an unfertilized egg cell
  The cell is now **enucleated** (no nucleus)
- Remove nucleus from a cell of another frog
- Insert egg cell nucleus into said cell
- If cell is in **blastula** stage — hollow ball of cells of an embryo, early embryo — then the cells divide into an adult frog, a clone of the frog that donated the egg cell nucleus
- If cell is past blastula — such as the later **gastrula** stage — the cells have already specialized, so they do not divide, and the embryo dies



Mammal Cloning

- More difficult
- Cells tend to be more specialized
- Nucleus transfer must be done before 8 cell stage of development
- Ensures nuclei are totipotent
- Needs **surrogate** — implanting an embryo into a surrogate and having the surrogate birth the offspring. No genetics from surrogate transfer.

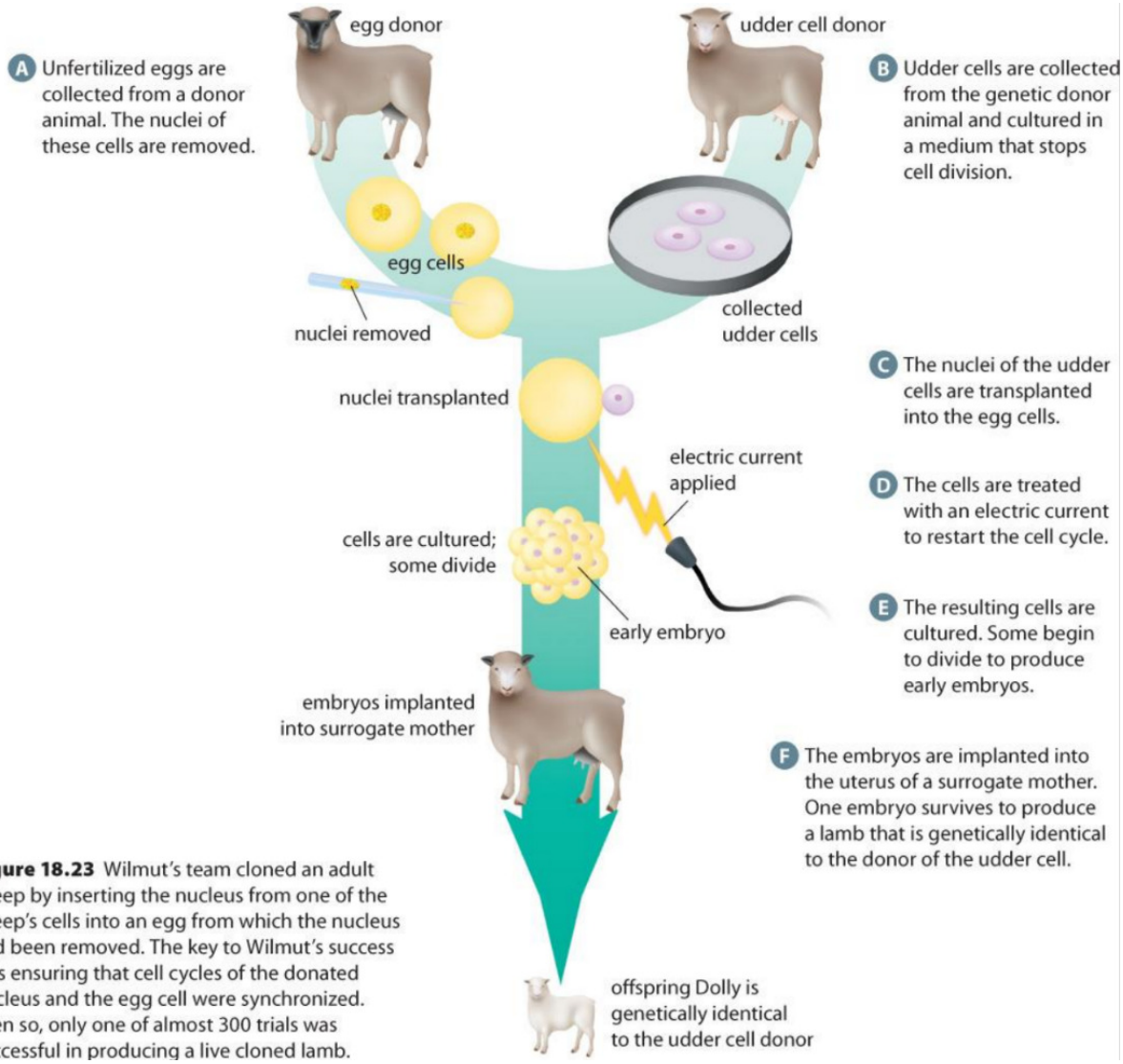


Figure 18.23 Wilmut's team cloned an adult sheep by inserting the nucleus from one of the sheep's cells into an egg from which the nucleus had been removed. The key to Wilmut's success was ensuring that cell cycles of the donated nucleus and the egg cell were synchronized. Even so, only one of almost 300 trials was successful in producing a live cloned lamb.

Cancer

- Rapid, uncontrollable growth of cells
- Divide faster than normal cells
- Some are very slow, some pause and return after many years
- Reproduce without directions from adjacent cells
- Cannot specialize — making them inefficient

Metastasis

- Cancer cells can dislodge from a tumor and move to another area
- Difficult to isolate source of cancer

Tumors

A mass of cancerous cells within otherwise normal tissue.

- **Benign Tumor**
  - If cancerous cells remain at site
  - Do not cause serious problems
  - Can be removed by surgery
- **Malignant Tumor**
  - If cancerous cells metastasize — dislodge & travel — and cause impairment of other organs
  - Unusual number of chromosomes

Causes

- x-rays
- chemical poisons
- asbestos
- fungi
- oncoviruses
- environmental factors (nature, e.g. diet)
- age
- inherited mutations

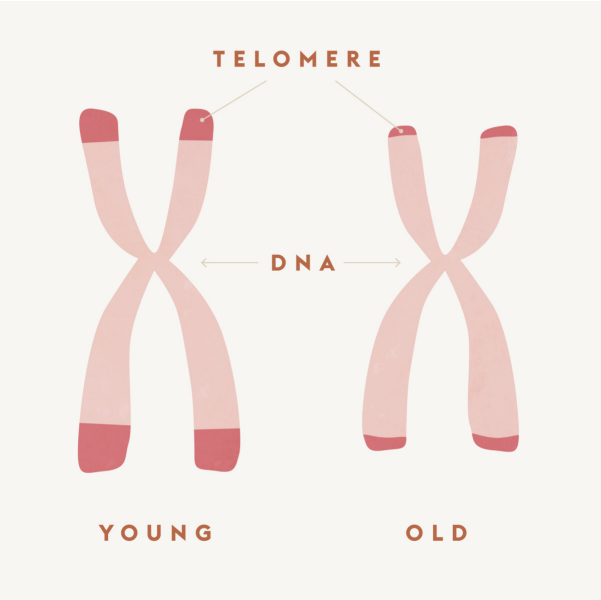
Methods of Identification

- x-rays
- cell biopsies

- infrared technology

Telomeres

- Caps at the end of chromosomes
- Reduce in length every cell cycle/division
- Clones — like Dolly — inherit their parents telomere length, shortening their life span compared to non-clones



Telomerase

- An enzyme that maintains telomere length, slowing cell death
- Not present in most normal cells
- Reactivated in cancer cells, explaining their immortality

(17.3) Sexual Cell Reproduction

Cons

- consumes a lot of energy
- infections
- only half of the genes are passed (not necessarily a con)
- males are deadbeat — contribute little to survival of offspring

Pros

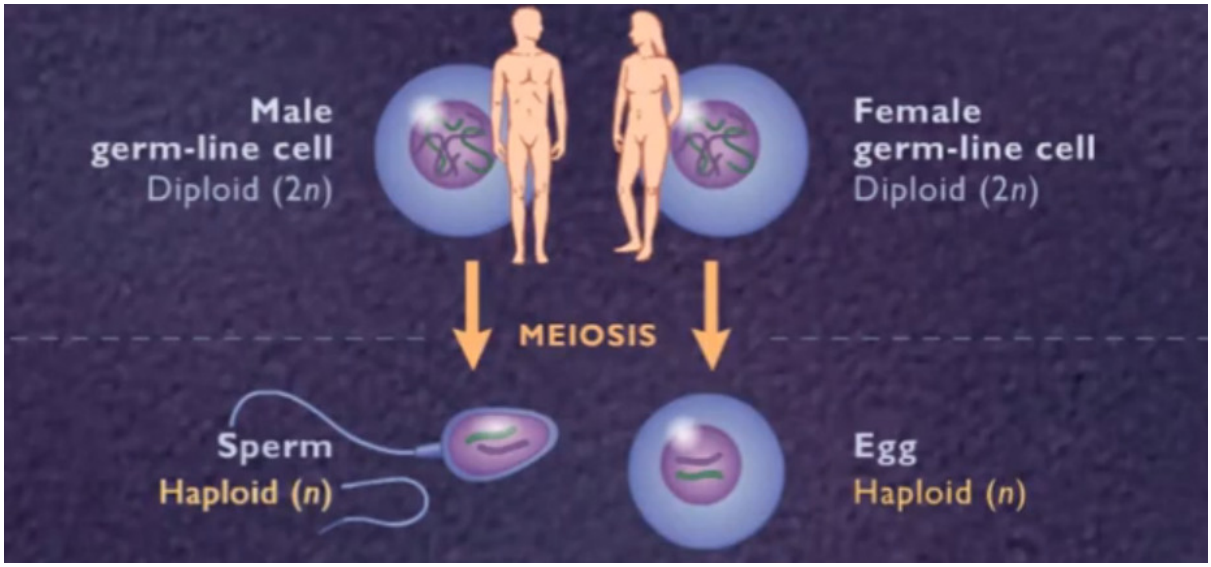
- Genetic diversity — more potential for survival if environmental conditions change.
- Genetic diversity comes from...
  - independent assortment — random shuffling and random order of genes in meiosis (metaphase I)
  - crossing-over in meiosis (prophase I)
  - random fertilization, combining genes of two separate individuals
- Two sets of chromosomes, so any damaged DNA has a backup, and a template to base DNA repairs off of

Meiosis

- **Gametes** = sex cells — ♀ ova/ovum (eggs) and ♂ sperm cells
- **Gonads** = reproductive organs — cells of ♀ ovaries and ♂ testes
- **Meiosis** = the process of forming gametes
- **Autosomes** = chromosomes not directly influenced by sex

- **Diploid** $(2n)$ = cell — such as somatic cells — with a typical # of chromosomes, such as 46 chromosomes in a human cell
- **Haploid** (n) = cell — such as gametes — with half the typical # of chromosomes, such as 23 chromosomes in a human gamete

Meiosis occurs in the germ cells of gonads.



Composition of Cells

Gametes

- 22 autosomes
- 1 sex chromosome
  - Ova can only have ♀X
  - Sperm can have either ♀X or ♂Y
- 23 total chromosomes

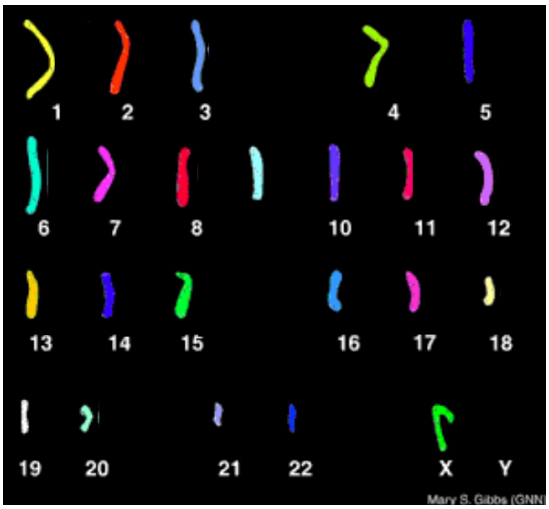
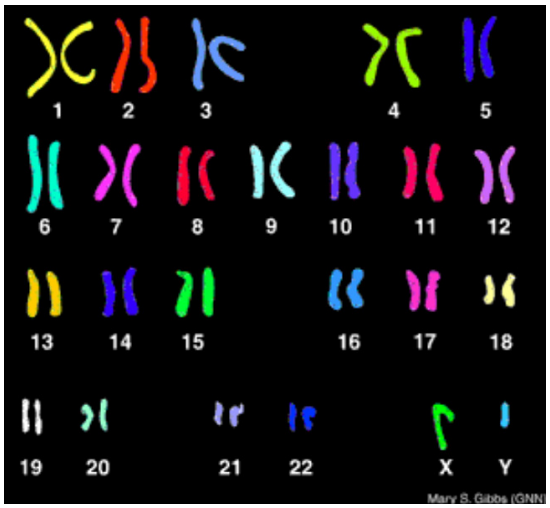


Figure 1: If this were a sperm cell, and it fertilized an egg, the baby would be female

Somatic

- 22 autosome pairs
- 2 sex chromosomes — either ♀XX or ♂XY
- 46 total chromosomes



Union

- 23 chromosome (haploid) sperm cell from ♂ male
- + 23 chromosome (haploid) egg cell from ♀ female
- = 46 chromosome (diploid) zygote or fertilized egg

Stages of Meiosis

Interphase (same as mitosis)

- Not splitting
- Important part: S phase — doubling 46 single chromosomes
- Ends up as 46 duplicated chromosomes (92 chromotids)

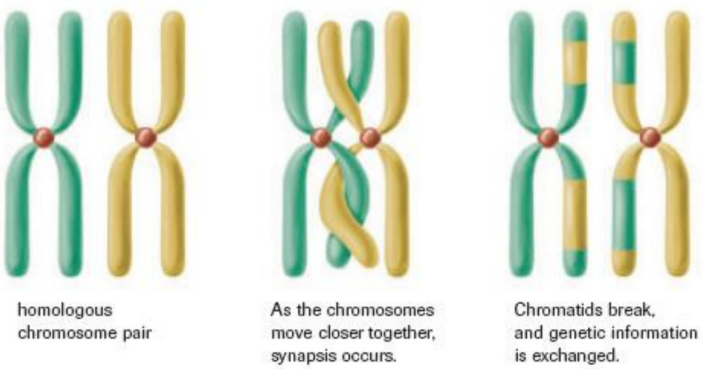
Meiosis I

Prophase I

- Same beginning as mitosis
  - Nuclear membrane dissolves
  - Centrioles move to opposite poles of cell, deploying spindle fibers
- **Homologous** = similar — such as shape, size, gene arrangement — but not identical
- Homologous chromosome pairs, one from the mother and one from the father, undergo **synapsis** — pairing side by side
- Sex cells from father and mother are homologous — also pair
- This forms a **tetrad** — 4 chromatids, homologous chromosome pair
- Chromosomes from the male and female shuffle around, as well as **crossover**

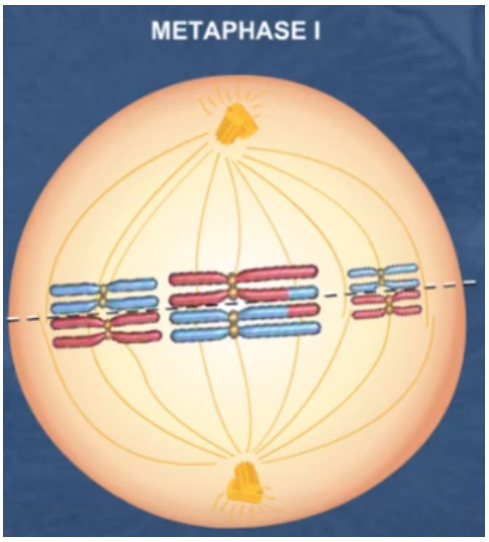
Cross-Over

- Inner chromatids of both chromosomes **cross-over** — genetic recombination, exchange genetic information
- Chromatids of both chromosomes are no longer sister chromatids after this point, not identical



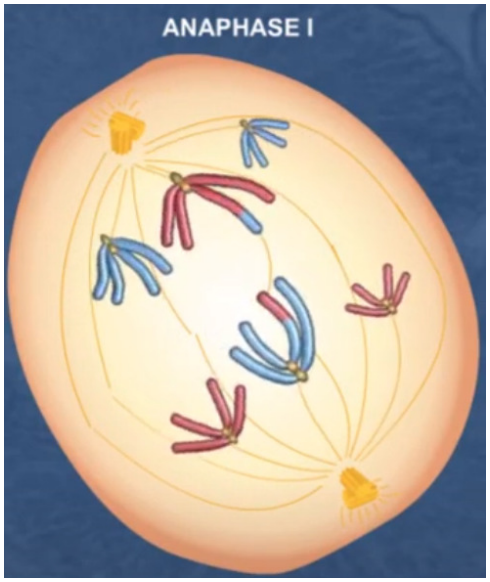
Metaphase I (mostly same as mitosis)

- Line up along equatorial plate, attach to spindle fibers
- Difference is instead of chromosomes lining up, tetrads line up



Anaphase I (mostly same as mitosis)

- Instead of splitting chromosomes, the homologous pairs are **segregated** (separated) and travel to opposite poles
- Diploid mother cell is now 2 haploid daughter cells



Telophase I (same as mitosis)

- The 2 cells are...
  - not identical to each other
  - not identical to parent cell
- Each chromosome remains double stranded

Meiosis II

Occurs at the same time in both of the daughter cells from Meiosis I.
No S phase.

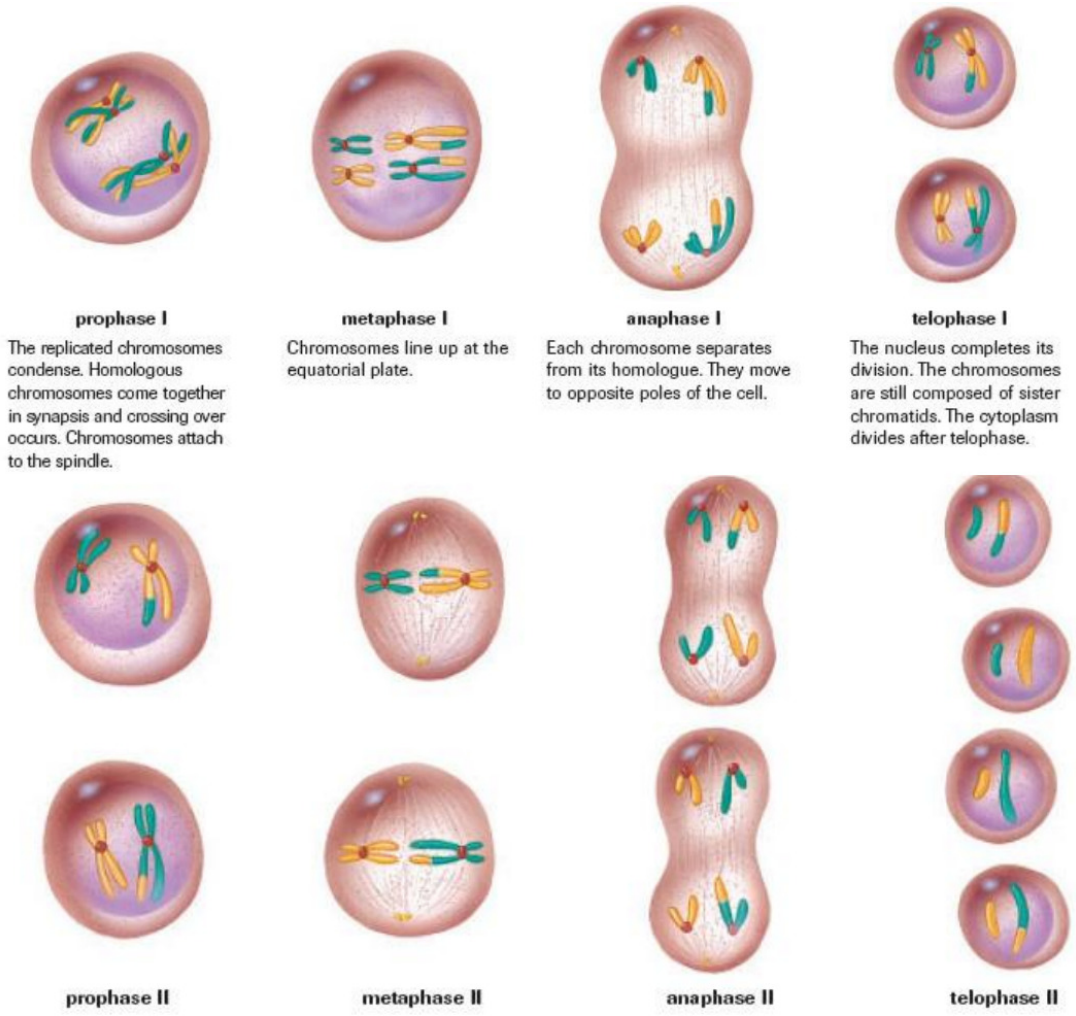
Same as Mitosis

The following stages occur identically to mitosis.

- Prophase II
- Metaphase II
- Anaphase II
- Telophase II

Conclusion

1 diploid mother somatic cell $\xrightarrow{\text{meiosis}}$ 4 haploid daughter gametes (sperm or egg)



prophase I
The replicated chromosomes condense. Homologous chromosomes come together in synapsis and crossing over occurs. Chromosomes attach to the spindle.

metaphase I
Chromosomes line up at the equatorial plate.

anaphase I
Each chromosome separates from its homologue. They move to opposite poles of the cell.

telophase I
The nucleus completes its division. The chromosomes are still composed of sister chromatids. The cytoplasm divides after telophase.

Mitosis vs. Meiosis

- Mitosis maintains ploidy level (# of chromosomes)
- Meiosis reduces ploidy level

- Meiosis only occurs in gonad cells
- Mitosis is far more common

- Mitosis
  - 1 division
  - daughter cells genetically identical to parent cell
  - produces 2 cells
  - 2n → 2n
  - produces cells for growth & repair
  - no crossing over
- Meiosis
  - 2 divisions
  - daughter cells genetically different from parent
  - produces 4 cells
  - 2n → 1n
  - produces gamete
  - crossing over

Differences Across Kingdoms

Reading a Life Cycle

You may be given the life cycle of a random species and need to identify whether a step is haploid or diplod. Just remember the following...

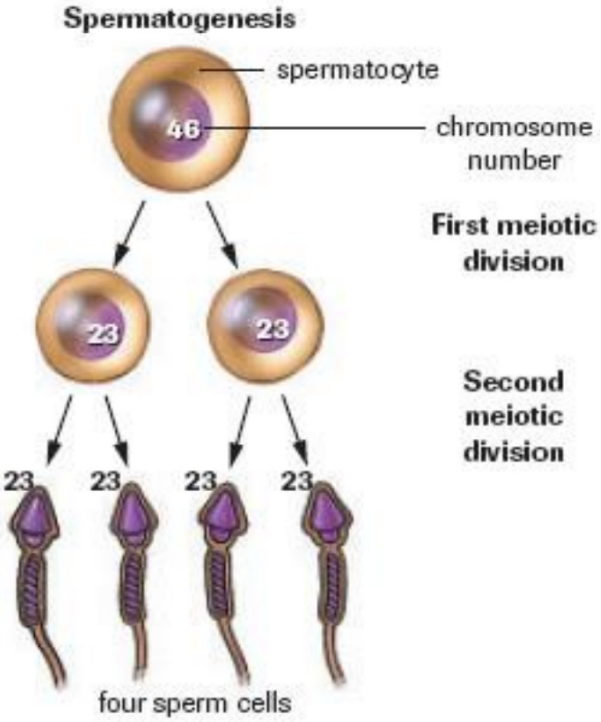
- Mitosis
  - $2n \longrightarrow 2n$
- Meiosis
  - $2n \longrightarrow n$
- Fertilization
  - $n \longrightarrow 2n$

Plant Sexual Reproduction

- **Alternation of Generations**
  - **Sporophyte** = non-sexual components of plant (e.g. pine tree, stem)
  - **Gametophyte** = sexual components of plant (e.g. pine cone, flower)
  - plant sporophyte ($2n$) and gametophyte (n) take turns reproducing each other
- Pollen are ♂ male sex cells
- ♀ Eggs are stored in a variety of locations
- Fertilization results in a seed
- Sporophyte (diploid, $2n$) $\longrightarrow$ Spores (haploid, n) $\longrightarrow$ Gametophyte (haploid, n)

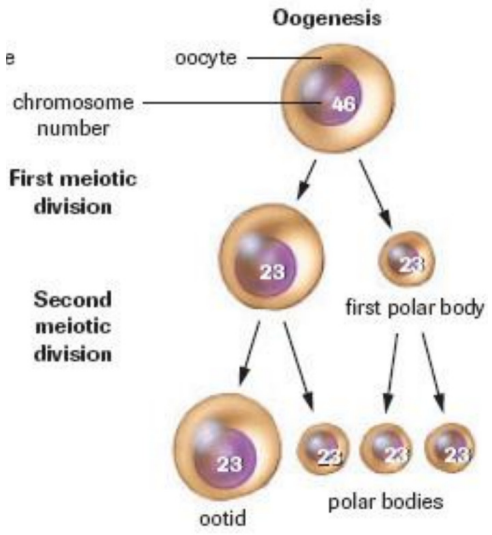
Development of ♂ Male and ♀ Female Gametes

- **Primary** = before meiosis I
- **Secondary** = after meiosis I
  - eggs pause during meiosis II — specifically metaphase II — to wait for sperm, needed to complete meiosis II
- **Gametogenesis** = formation of gametes during meiosis
- **Spermatogenesis** = formation of sperm cells
- **Spermatocyte** = a diploid cell that undergoes meiosis to become 4 sperm cells
  - Capable of many mitotic divisons before meiosis
  - Explains males being able to produce 1 billion sperm per day

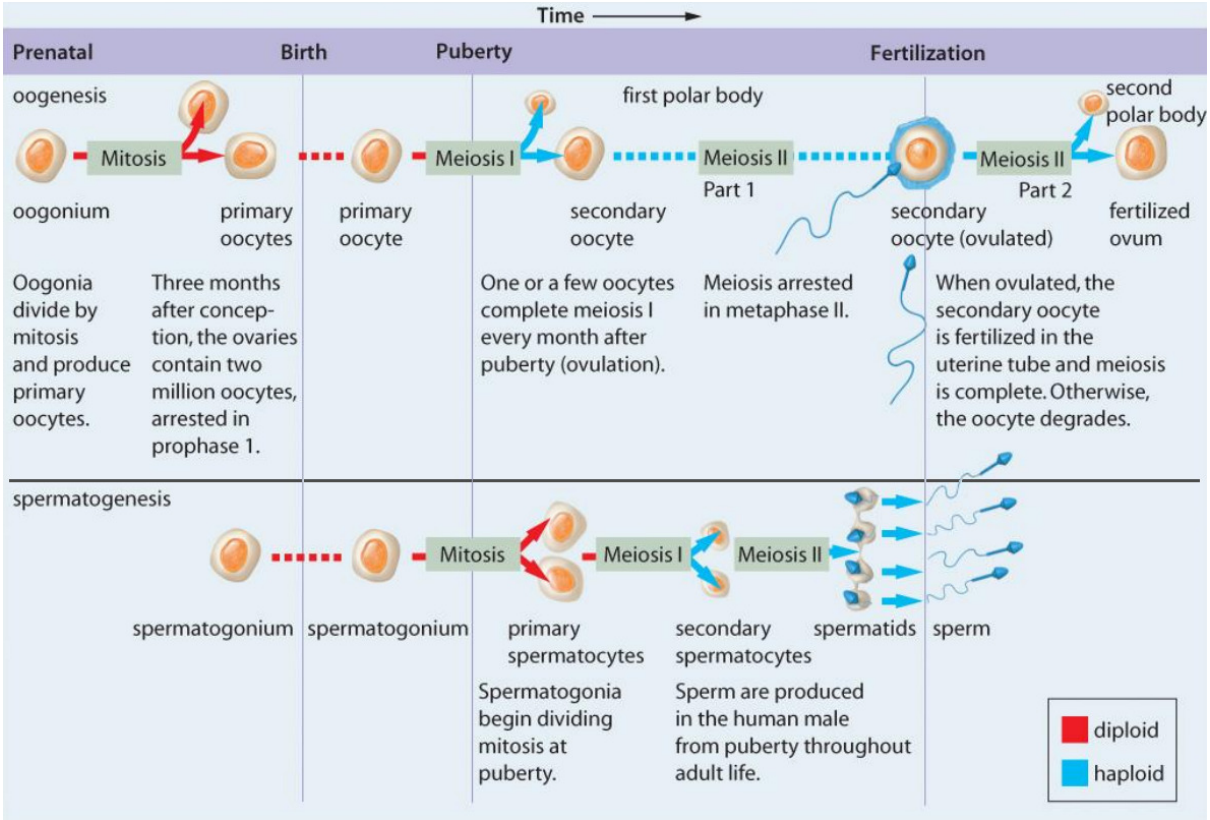


Oogenesis

- Cytoplasm of female gametes (eggs) is not divided equally after every division
- **Ootid** (aka. oocyte) = The one daughter cell that recieves the most cytoplasm
- **Polar Body** = The other daughter cells die, their nutrients absorbed
- Only one egg is viable for fertilization every division



Immature $\longrightarrow$ Mature



(17.4) Nondisjunctions

Also known as **abnormal meiosis**.

- Occurs when 2 homologous chromosomes move to the same pole
- Occurs during anaphase in mitosis, meiosis I, or meiosis II (test question)
- A cell will be missing a chromosome, and another will have an extra
- Cells with too much or too little genetic information will not function correctly

Anaphase I & II

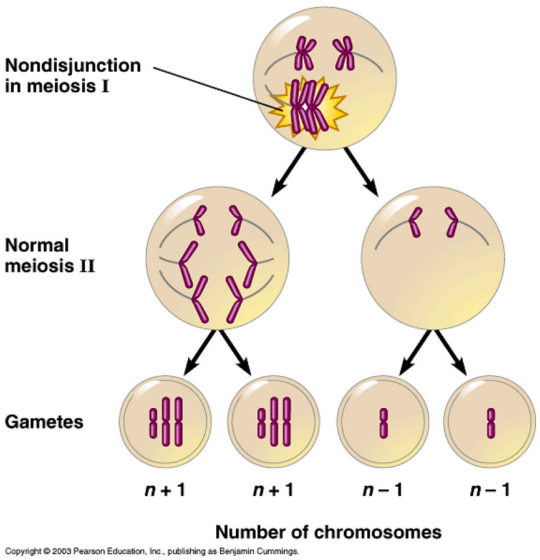
Nondisjunction can occur in either anaphase I or anaphase II. The difference is...

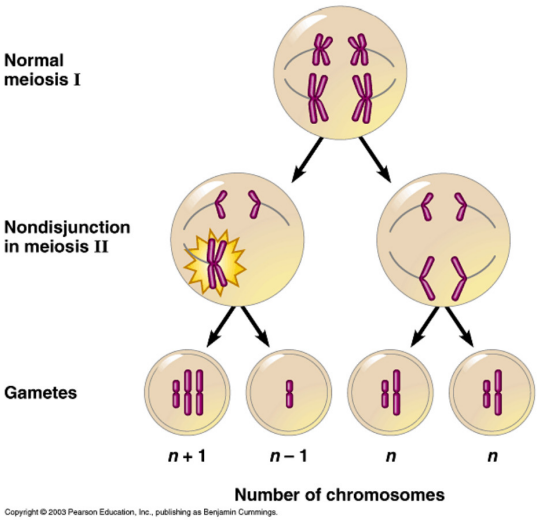
- **Nondisjunction in anaphase I**

  2 cells have too many chromosomes, 2 cells have too little chromosomes
- **Nondisjunction in anaphase II**

  1 cell has too many chromosomes, 1 cell has too little chromosomes

This is a test question.





Terms

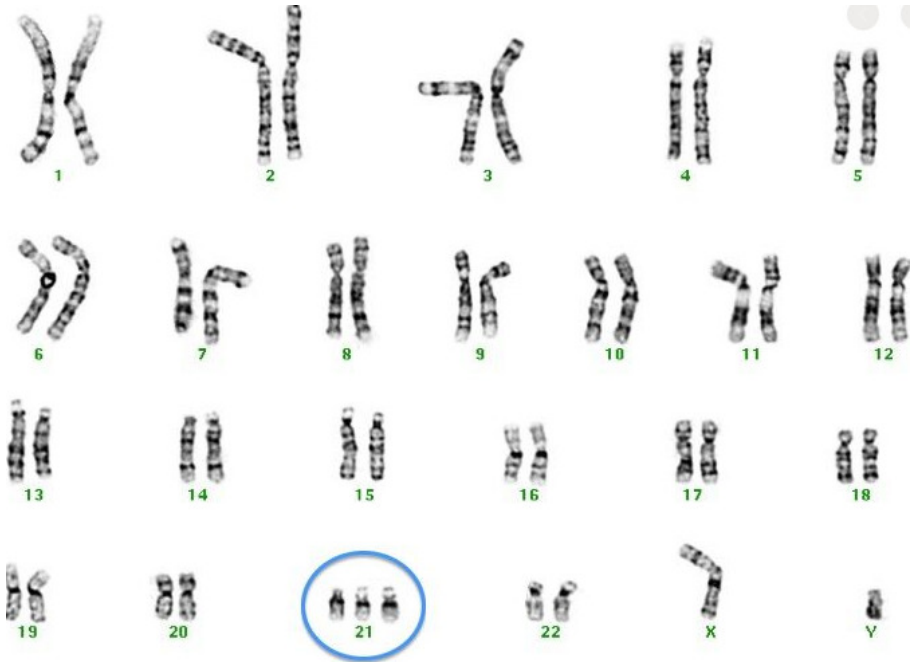
- **Karyotype chart** = A picture of chromosomes, arranged in homologous pairs
- **Polyploidy** = An organism with > 2 complete sets of chromosomes
  - **Triploidy** $(3n)$ = may result from abnormally diploid $(2n)$ egg fertilized by normal (n) sperm, or vice versa
  - **Tetraploidy** $(4n)$ = doesn't occur in humans, failure of diploid zygote to divide after duplicating chromosomes following mitosis
  - **Aneuploidy** = all cells of the body contain abnormal # of chromosomes
- **Trisomy** = fertilized egg with 3 # of a chromosome (normally 2)
  normal gamete (23 pairs) + abnormal gamete (24 pairs), 47 chromosomes total
- **Monosomy** = fertilized egg with 1 # of a chromosome (normally 2)
  normal gamete (23 pairs) + abnormal gamete (22 pairs), 45 chromosomes total

Syndromes

Down Syndrome

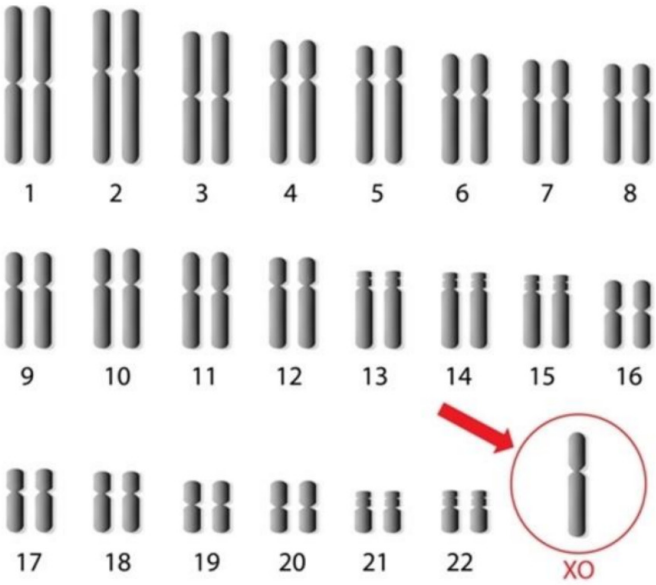
aka. Trisomy 21

- Extra chromosome in pair #21 (trisomic disorder)
- Causes...
  - mentally challenged
  - round, full face
  - enlarged, creased tongue
  - short
  - large forehead



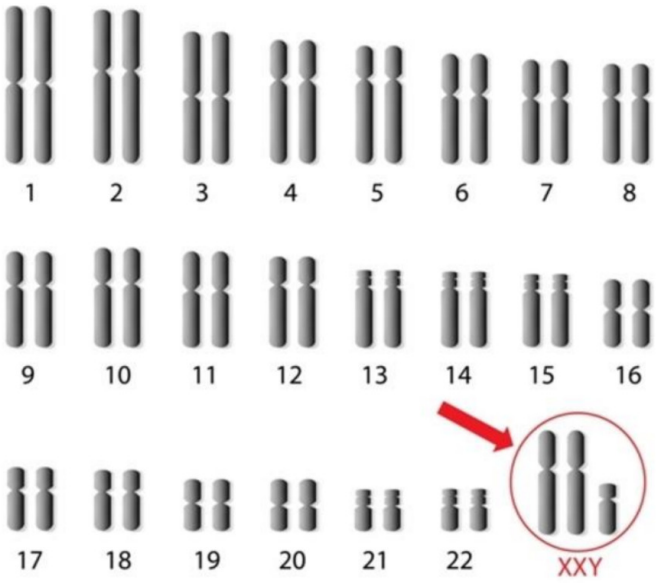
Turner's Syndrome

- Female with a single ♀X chromosome (instead of ♀XX) (monosomic disorder)
- Causes...
  - no sexual development
  - short
  - thick, widened necks



Klinefelter Syndrome

- Male with an extra ♀X chromosome (XXY instead of XY) (trisomic disorder)
- Causes...
  - high estrogen
  - sterility



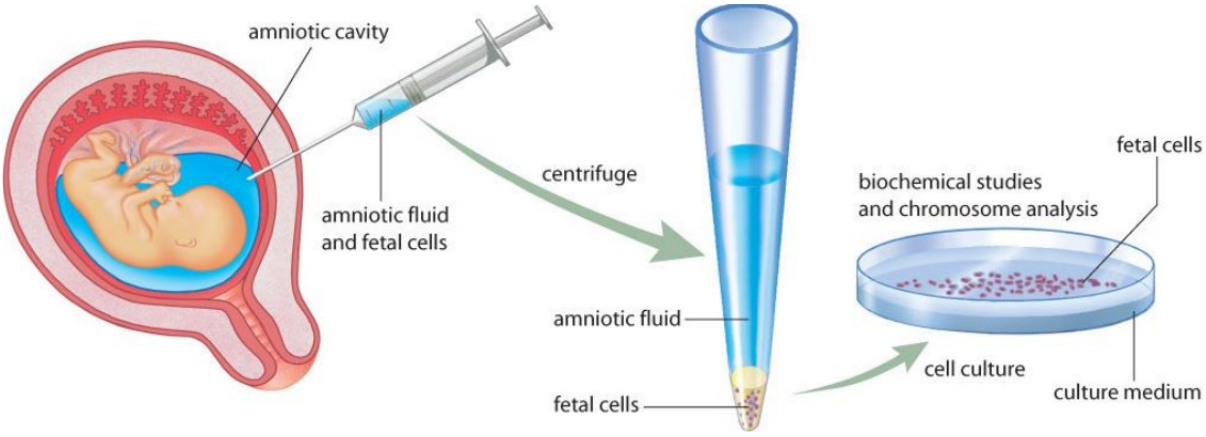
Teratogenic Compounds

- Chemicals that cause abnormalities in embryos
- drugs (e.g. alcohol), infectious agents (viruses), radiation

Diagnosis of Fetus

Amniocentesis

- Use of a syringe to draw fluid from sac surrounding fetus
- Analysis can identify disorders, down syndrome, and sex
- Amniotic fluid contains not a lot of cells from the fetus, so results take a while
- **Ultrasound** = used to locate position of fetus in womb



Chorionic Villus Sampling (CVS)

- Drawing cells from outer membrane surrounding embryo
- Can be done earlier and results quicker than amniocentesis

(20.1) DNA

- Deoxyribonucleic Acid
- Carrier of genetic info and instructions that ensure continuity of life (common diploma term)
- Regulates production of cell protein
- Only molecule that can duplicate itself

Names

- **Franklin** = female who discovered it, or something
- **Watson** & **Crick** = guys who yoinked it and won the nobel prize
- Watson & Crick proposed double helix structure

Basic Units

- **Nucleotide** = basic unit of DNA
- Comprised of...
  - phosphates
  - deoxyribose sugars
  - nitrogen-containing bases

Nitrogen-Containing Bases

- A = **Adenine**
- T = **Thymine**
- G = **Guanine**
- C = **Cytosine**

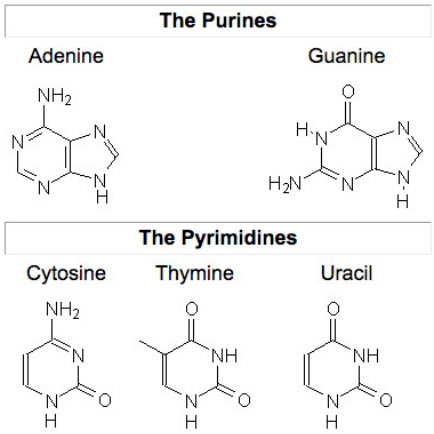
- The following always pair with one another in DNA, so they are in equal quantities
  - # of A = # of T
  - # of G = # of C

Structure

- Double helix (twisted ladder)
- Sugar and phosphate molecules form "backbone/spine" of ladder
- N bases form rungs of ladder
- N bases of different spines are bonded together via weak hydrogen bonds

Complementary Pairs

- N bases are always paired purine + pyrimidine
- **Purine** = 2 ring structure (A, G)
- **Pyrimidine** = 1 ring structure (C, T, U)



Anti-Parallel

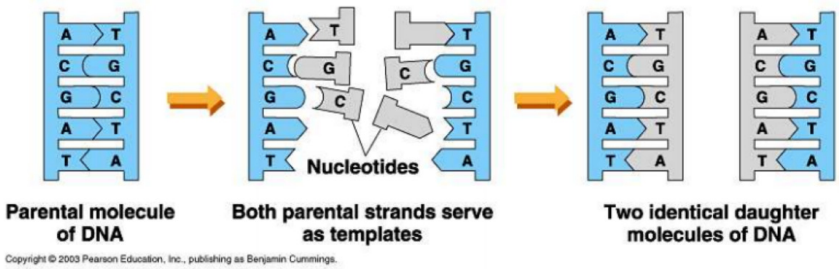
- The strands of the double helix are parallel
- However, they run in opposite directions, upside-down to one another
- Strands have positive and negative ends, upside-down to balance

DNA Replication

- DNA is duplicated during S phase interphase
- Process is semiconservative — one strand is duplicated, the other is the old one

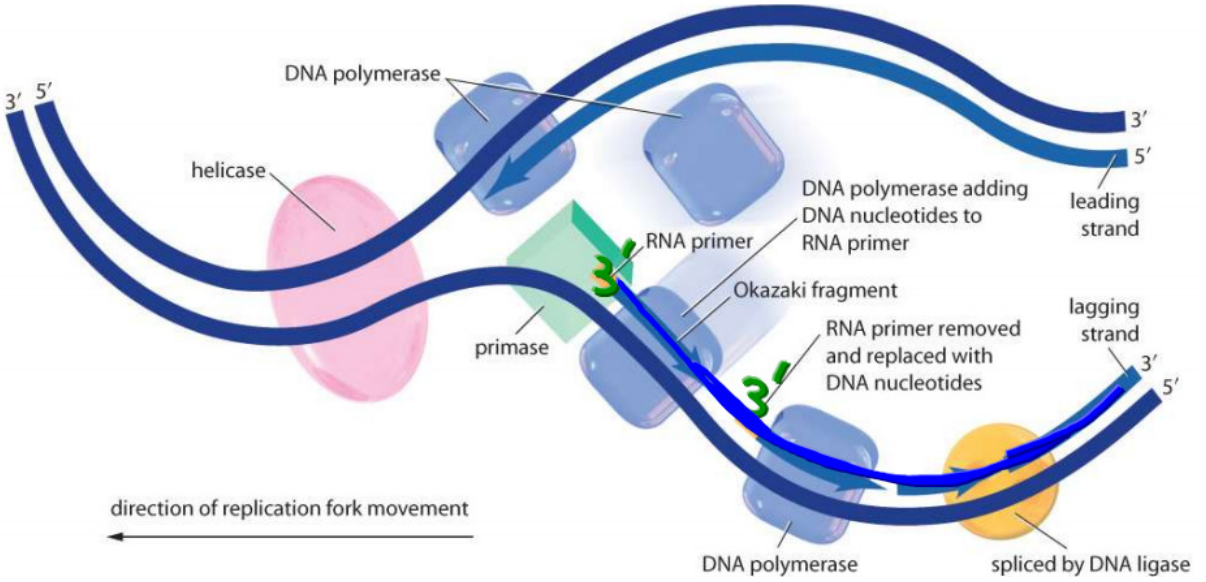
Steps (simplified)

1. Hydrogen bonds break. DNA helix unzips
2. Each strand acts as a template to build the complementary strand
3. Errors are repaired
4. Two identical copies of DNA in the end



Steps

1. **DNA helicase** enzyme
    - Unwinds helix by breaking hydrogen bonds between complementary base pairs
    - The point where the two strands separate is called the **replication fork**
2. **DNA polymerase III** enzyme
    - Links together free nucleotides (DNA from the food you eat) that have bases complementary to the template strand
3. **DNA ligase** enzyme
    - The two strands of DNA from the split are treated differently
    - **Leading** vs **lagging** strand
    - Leading strand written continuously by DNA polymerase III, ligase not needed
    - Lagging strand written in chunks
    - Ligase glues together the sugar-phosphate backbone and DNA fragments/chunks in lagging strands, filling in the gaps
4. **DNA polymerase I & III** enzyme
    - Uncomplimentary N bases may become paired
    - These enzymes proofread the DNA and fix any errors/mutations from hazardous chemicals or radiation

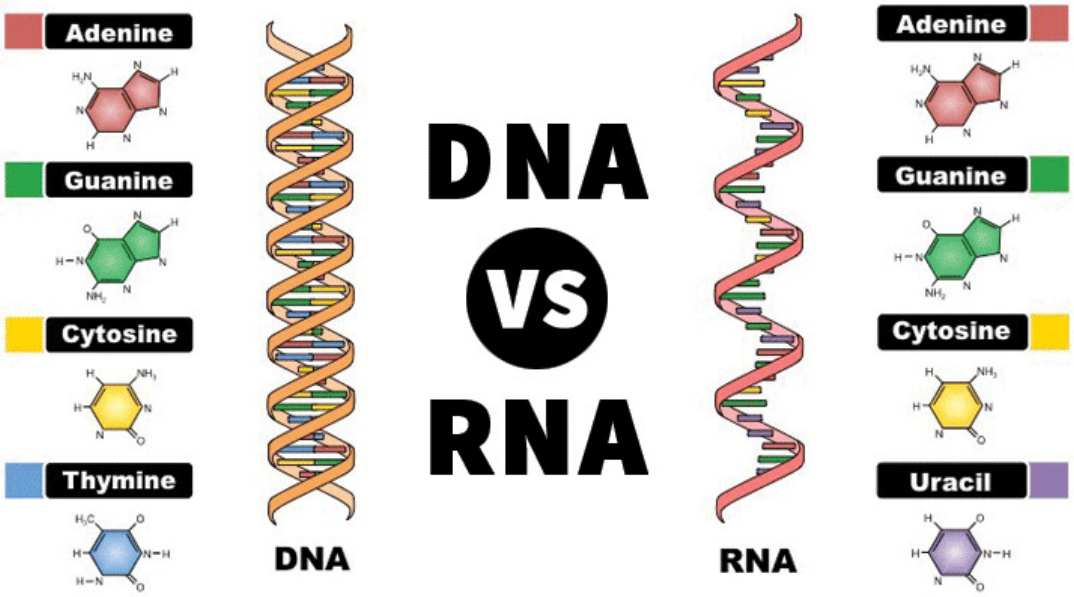


(20.2) Protein Synthesis

- Sequence of N bases of DNA determines which proteins are made & the activities of proteins
- DNA too large to leave nucleus during protein synthesis
- **Messenger RNA**, **mRNA**, is used instead
  - Reads DNA code and carries it to ribosomes

RNA vs. DNA

- RNA has a ribose sugar, instead of a deoxyribose sugar
- RNA has no thymine; **uracil** (U) in its place
- RNA is single-stranded, DNA is double-stranded



Steps

Transcription

Occurs in nucleus.

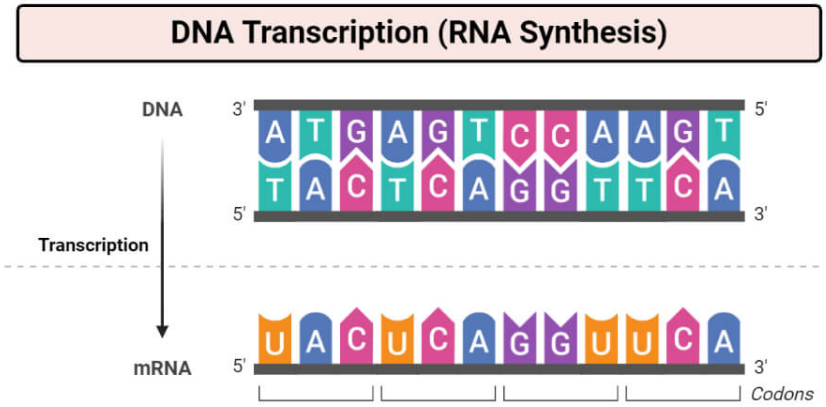
Initiation

- RNA polymerase binds to promoter sequence (not transcribed) on DNA
- RNA polymerase allows for nucleotides to attach along mRNA

Elongation

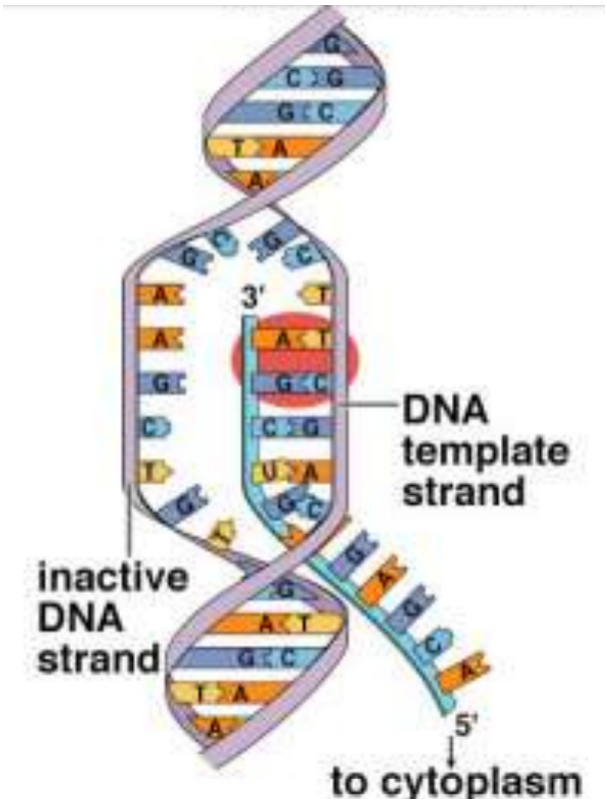
- DNA unzips
- mRNA reads single strand from the DNA, known as the **template strnad**

- mRNA finds complementary pair for each nucleotide on template strand
  - DNA cytosine $\longleftrightarrow$ mRNA guanine
  - DNA thymine $\longrightarrow$ mRNA adenine
  - DNA adenine $\longrightarrow$ mRNA uracil (RNA has no thymine, uracil instead)
- mRNA joins complementary nucleotides to a long chain



Termination

- mRNA moves away from DNA, disconnecting the chain it made
- 2 strands of original DNA rejoin
- Single-stranded mRNA molecule moves through nuclear membrane, carrying N base code to ribosomes in cytoplasm



Translation

Occurs in cytoplasm.

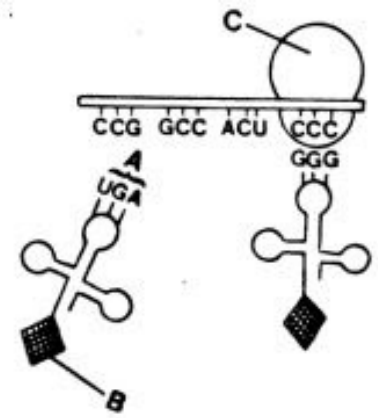


Figure 2: A = **Anticodon**, B = **Amino acid**, C = **Ribosome**

Initiation

- mRNA attaches itself to ribosomes like a ribbon
- **Codon** = 3 nucleotides that are code for an amino acid
- **Initiator codon** turns on protein synthesis (**AUG**, methionine, always at beginning)
- mRNA codons
  - Codons — blocks of 3 nucleotides — are decoded into a sequence of amino acids
  - Nucleotide sequence to amino acid conversion table is located on page 3 on the data sheet

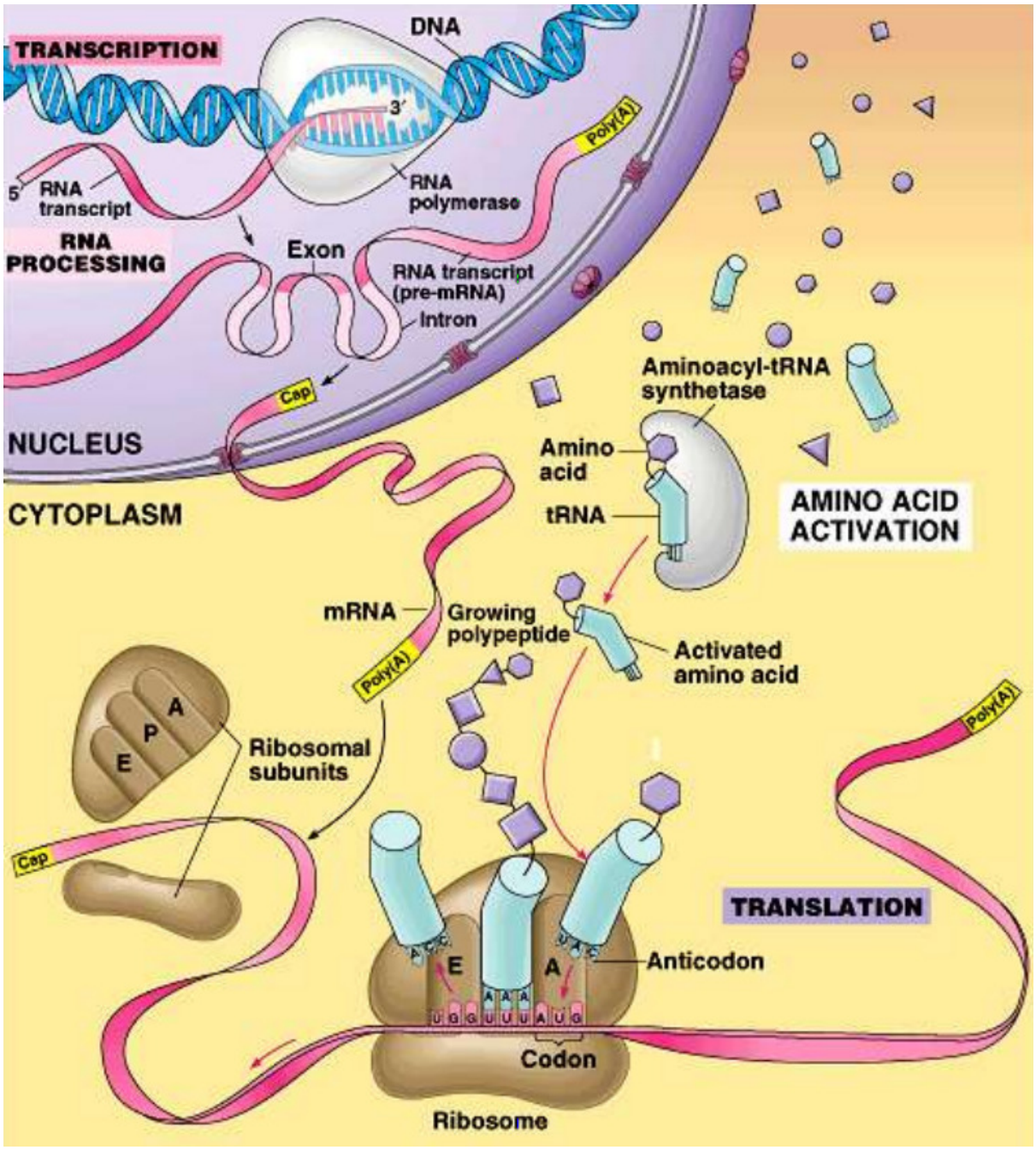
Elongation

- **Transfer RNA** (tRNA) picks up amino acids in cytoplasm and sends to mRNA
- mRNA codon and tRNA anticodon are complementary
- tRNA molecule is T-shaped
- Amino acids — brought by tRNA — are fused into long-chain proteins at ribosome on the top of each tRNA that brings each amino acid
- Amino acids are bonded together by **ribosomal RNA** (rRNA)

Termination

- **Terminator codon** turns synthesis off, always at the end
- Terminator codon can be either...
  - **UAA**
  - **UGA**
  - **UAG**

Central Dogma



- **Central Dogma** = main idea, diploma term
- A DNA sequence encodes an RNA sequence that encodes protein

Code Format Summary

- DNA $\longleftarrow$(complementary, $\mathrm{T} \longleftrightarrow \mathrm{U}$)$\longrightarrow$ mRNA $\longleftarrow$(complementary)$\longrightarrow$ tRNA
- DNA $\longleftarrow(\mathrm{T} \longleftrightarrow \mathrm{U})\longrightarrow$ tRNA

(20.3) Biotechnology

Genetically Modified Organisms (GMO)

- **Recombinant DNA** = piece of DNA composed of sequences from 2+ different sources
- **Genetic transformation** = introduction and expression of foreign DNA in a living organism

Steps

- **Restriction endonucleases/enzymes** = cut DNA at a specific base, specific recognition site
- **Recognition site** = 4-8 base pairs long, restriction enzyme scans DNA until recognition site (e.g. Eco R1)
- Cuts on both strands, so one strand will be longer than the other; this "overhang" is called a **sticky end** (diploma term)

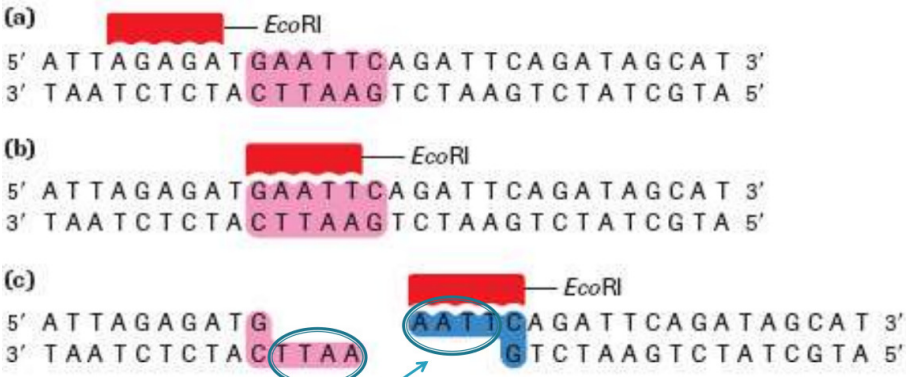
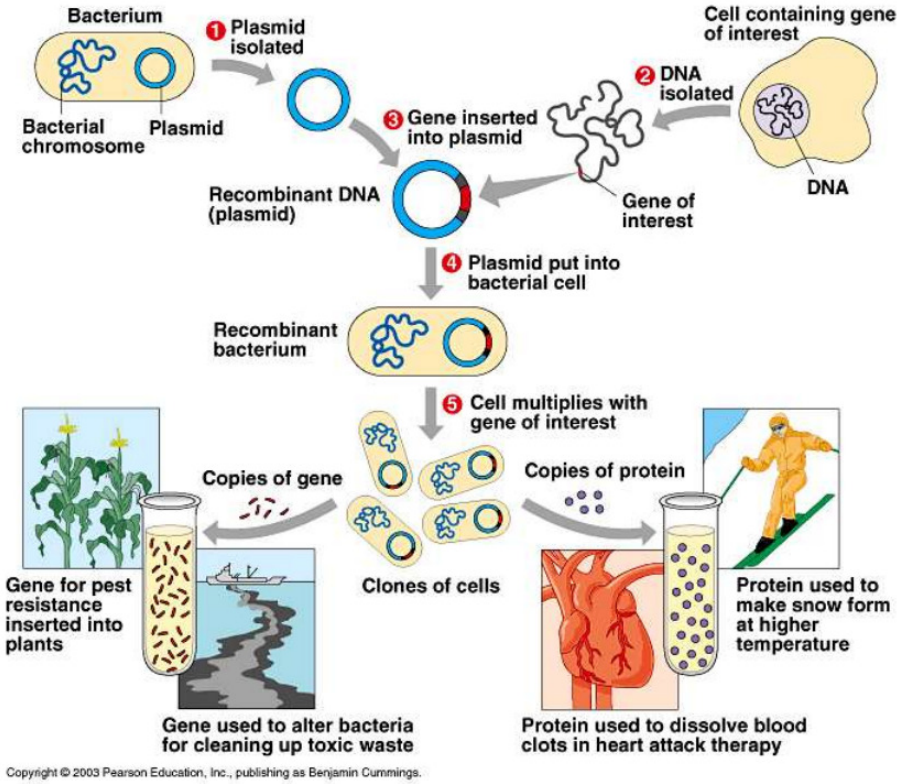


Figure 3: "Sticky ends" circled

- **anneal** = glue/join (diploma term)
- **DNA ligase** = foreign DNA is inserted between sticky ends and anneled to them
- **Methylases**
  - Enzymes that can modify a restriction enzyme recognition site
  - Add methyl group to one of bases in site to protect its own DNA from digestion by its own restriction enzymes

Transgenic Bacteria

- Annealing bacteria plasmids in order to force them to do our bidding
- Most popular example is annealing insulin producing DNA into bacteria plasmids
- This forces the bacteria to produce insulin, rather than harvesting it from animals
- Other examples include bacteria that eat toxic waste

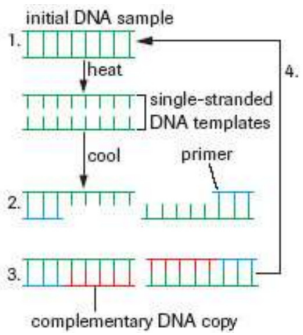


Polymerase Chain Reaction (PCR) with Taq DNA Polymerase

- Allows billions of copies from small quantities of DNA
- Stable at high temperatures

Steps (simplified)

- DNA heated to break hydrogen bonds
- Cooled, primers form hydrogen bonds with DNA templates
- Taq polymerase creates new DNA strand using template via complementary base pairing, starting at each primer
- Repeat for more DNA copies



DNA Fingerprint Test

At the Alberta level, all you would be tested on is...

- Matching the black bands — called **RFLP**s — of the DNA samples
- Identify which sample is more similar

(20.4) Mutations

- Changes in a sequence of DNA
- **Mutagenic agents** = things that alter DNA
  - cosmic rays
  - x-rays
  - UV radiation
  - chemicals

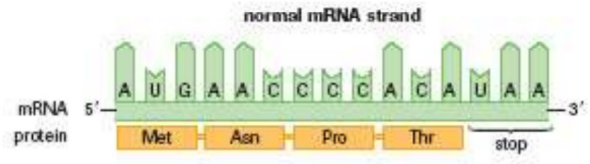
  Especially harmful during 1st trimester of pregnancy
- Gamete mutations lead to permanent change in offspring characteristics

Classes

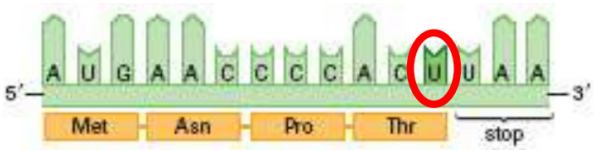
- **Beneficial mutations** = selective advantage, tends to become more common over time, leads to evolutionary change
- **Harmful mutations** = reduces an individual's fitness, tends to be selected against, occurs at low rates
- **Neutral mutations** = no benefit nor cost, not acted on by natural selection

Point Mutations

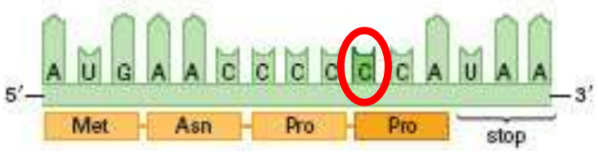
Changes a single base pair in DNA.



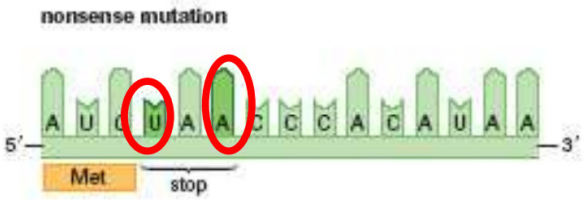
- **Silent mutatation** = no effect; doesn't change amino acid coded for



- **Missense mutatation** = changes one amino acid coded for; e.g. sickle-cell anemia

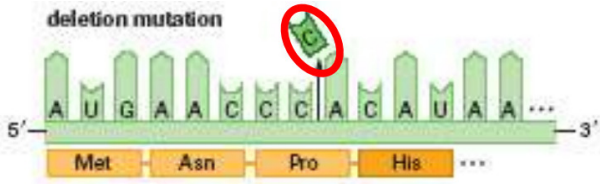


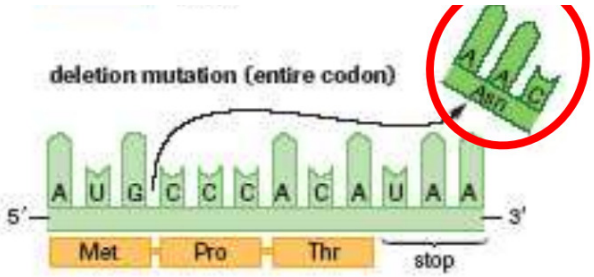
- **Nonsense mutatation** = converts an amino acid codon into a stop codon, part of protein may be digested by cell proteases, often lethal



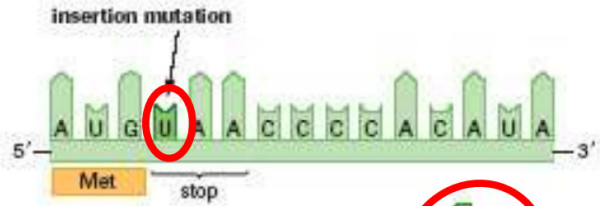
Gene Mutations

- Changes the amino acids specified by DNA sequence
- May involve 1 or more base pairs
- Both cause **frameshift mutations**, shifting all the nucleotides, causing completely different codons to be read
- **Deletion mutation** = 1 or more nucleotides removed from DNA sequence





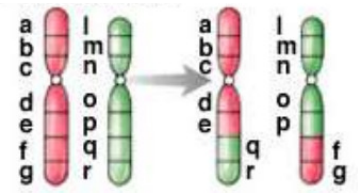
- **Insertion mutation** = extra nucleotide inserted into DNA



Chromosomal Mutations

Involves large segments of DNA

- **Translocation** = relocation of groups of base pairs from 1 part of a genome to another



- **Inversion** = section of chromosome reversed in orientation

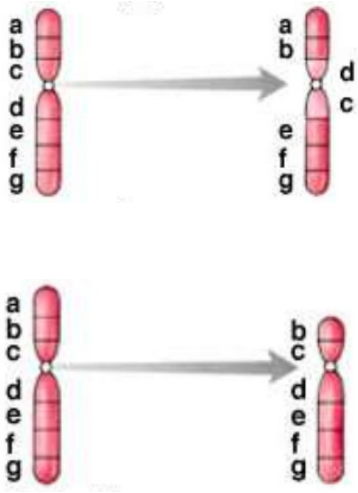


Figure 4: Deletion

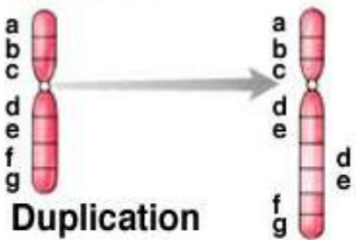


Figure 5: Duplication

Mutation Examples

- **Hemophilia** = absence of protein needed for blood clotting
- **Cystic fibrosis** = deletion; inability to produce protein that regulates Cl^- channels, lung secretions thick and block airways

Causes

- **Spontaneous mutations** = errors made in DNA replication, DNA polymerase I, results in point mutation
- Mutagenic agents

Oncogenes

- 'onco-' = cancer
- Cancer-causing genes
- Present in normal strands of DNA
- Regulator gene keeps oncogenes turned off
- Translocation allows oncogene to turn itself on

Mitochondrial DNA (mtDNA)

- identical to one's mother's mtDNA
- **Eve Project** = tracing mutations in mtDNA, shows ancestry

IB TOPICS

In your booklet. :)