BIOL206 lecture notes
Genetics
Lecture 1: Introduction

Genetics
- The study of genes – hereditary material

Genetics in Agriculture: Selective Breeding and GMO's
- Food security

Genetics in Medicine
- Mutant/deleterious alleles
- Genetic applications – disease treatment, disease risk
- Genetic counsellors

Genetics role in society
- Economic (biotech, pharmaceuticals), legal (paternity, forensics), philosophical

Areas being covered
- Classical Genetics (Mendelian)
    o Analysis of outcomes of crosses between strains of organisms
    o Structure and behaviour of chromosomes
- Molecular Genetics
    o Replication, expression, mutation of genes at the molecular level
    o Study of DNA sequences and manipulation of DNA molecules
- Environmental/Evolutionary Genetics
    o Different alleles of genes
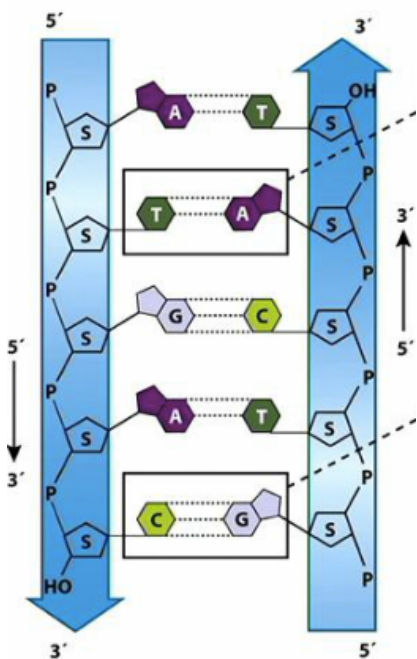   o Ecological genetics based on analysing allele and genotype frequencies

Lecture 2: Revision I

Structures of DNA/RNA
- DNA – double stranded, A-T, C-G
- RNA – single stranded, A-U, C-G
- A-T = 2 H bonds, C-G = 3 H bonds

Polynucleotide chain

Opposite polarity of the two strands

Chromosomes
- Linear end-to-end arrangement of DNA
- 2 main activities:
    o Transmit info from cell→cell and from one generation→generation
    o Express the info to control cellular function and development
- 2 sister chromatids – identical copies of parental chromosomes, 2 DNA molecules
- Homologous Chromosomes – occur in pairs and are similar in shape/size, one being inherited from the female parent and the other from male parent
    o Same genes
- Metacentric – centromere in the middle
- Acrocentric – centromere towards one end
- Telocentric – centromere at the end

DNA Packaging
- DNA → nucleosomes → chromatin → chromosome
- 2nm double stranded DNA molecule, 11nm nucleosome, 30nm chromatin

Gene, Locus, Allele
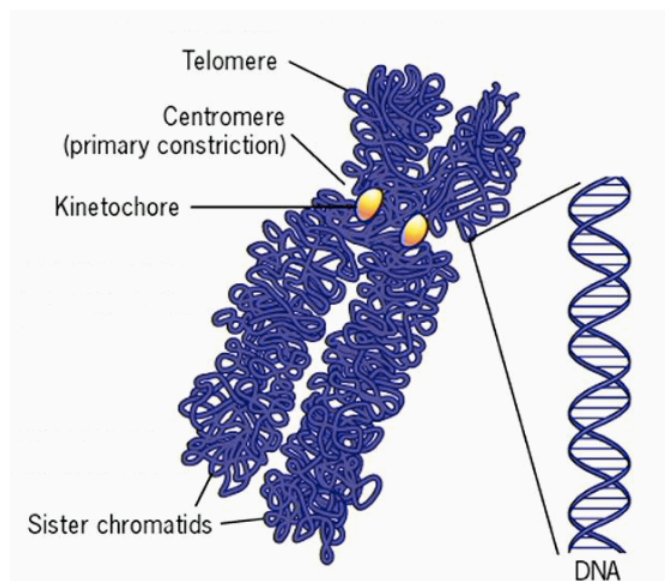
BIOL206 lecture notes
Genetics
- ABO locus (blood type) – IAIA or IAi = A, IBIB or IBi = B, IAIB = AB, ii = O

Pedigree
- The first generation of descent from a given mating – F1 = the first generation of sons and daughters
- The second generation produced by the F1 mating with each other – inbred grandchildren of a given mating (controlled genetic experiment)

Punnet Squares
- When looking at 2+ (unlinked) genes:
  - 1. Work out the probabilities for the 1st gene by itself
  - 2. Work out the probabilities for the 2nd gene
  - 3. Continue looking at each gene separately until the probabilities for each gene has been calculated
  - 4. Multiply the probabilities

Probability
- Product rule (independent events) – AND = MULTIPLY
- Sum rule (mutually exclusive events) – OR = ADD

$$\text{Probability } (P) = \frac{\text{no. times event is expected}}{\text{no. opportunities for event}}$$

Lecture 3: Revision II – Sex and Reproduction
Basic Definitions
- Somatic – body cell, genes wont be passed onto next generation
- Germ cell – reproductive cell, fertilisation
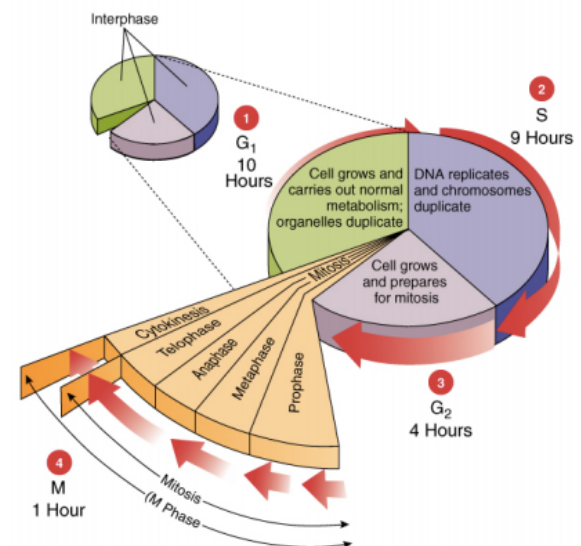- Gamete – A mature male or female reproductive cell (haploid)

Reproduction vs. Sex
- Reproduction = 1 cell → 2
- Sex = 2 cells → 1
  - Unnecessary for reproduction, slows
  - What is sex? Cyclic alternation of fusion of haploid gametes → diploid zygote and regeneration of haploid cells from diploid (meiosis)
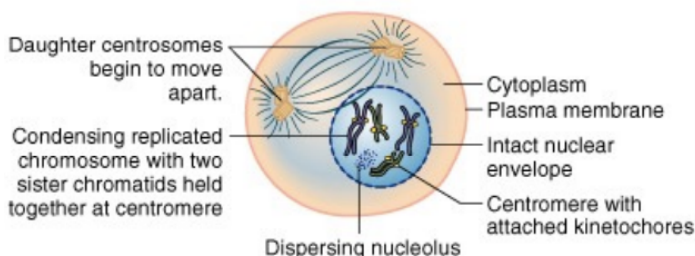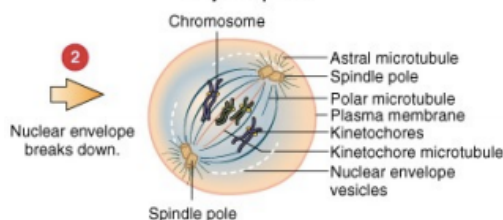 - Why sex? Increases genetic variation (Natural Selection)

The cell cycle

Mitosis
- Cell division to produce 2 genetically identical daughter cells (same # of chromosomes)
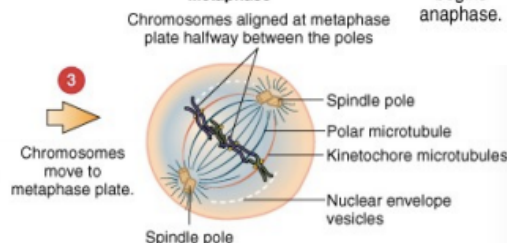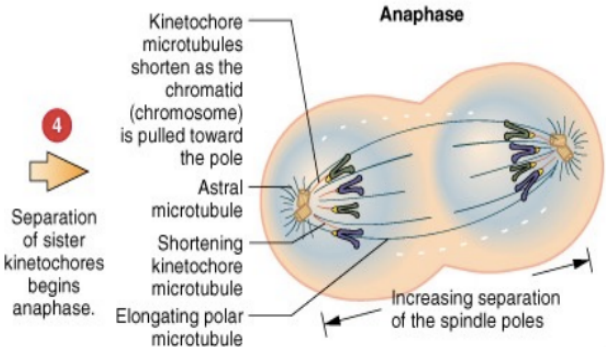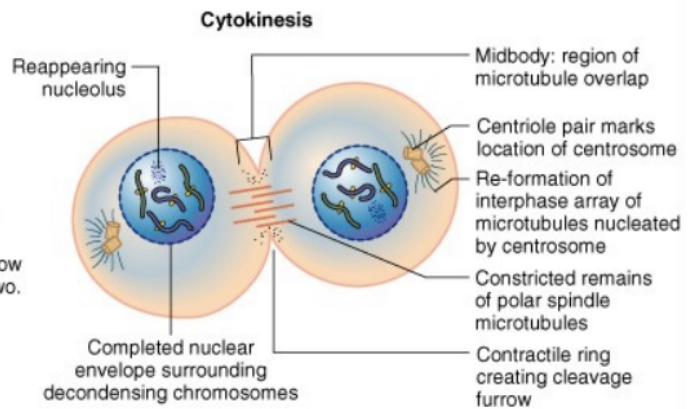- Prophase → metaphase → anaphase → Telophase/cytokinesis)

Telophase

Polar microtubules

Decondensing chromosomes without kinetochore microtubules

Nuclear envelope re-forming

6

Cleavage furrow splits cell in two.

Cytokinesis

Reappearing nucleolus

Midbody: region of microtubule overlap

Centriole pair marks location of centrosome

Re-formation of interphase array of microtubules nucleated by centrosome

Constricted remains of polar spindle microtubules

Contractile ring creating cleavage furrow

Completed nuclear envelope surrounding decondensing chromosomes
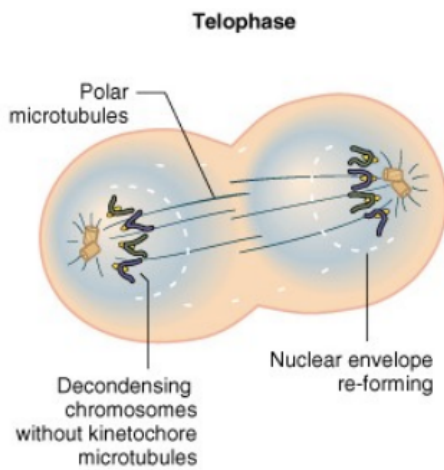
Meiosis

- Chromosome # becomes reduced to half the diploid (2n) or somatic #
- Changes genetic info to increase diversity
- 2 successive divisions:
    - o 1. Reductional division: chromosome # is halved
    - o 2. Equational division: sister chromatids separate
- Meiosis 1:
    - o Prophase I
        - ▪ Chromosomes condense, each chromosome has 2 sister chromatids
        - ▪ Synapsis of homologous chromosomes, synaptonemal complex
            - • Synaptonemal complex = DNA and protein between homologous chromosomes in synapsis/crossing over, recombination nodules have enzymes for crossing over
        - ▪ Chromosomes condense further, bivalent, crossing over occurs
            - • Crossing over causes recombination – recombinant gametes, new combination of alleles/genes = genetic variation
        - ▪ Paired chromosomes separate slightly but are in contact as chiasmata
            - • Chiasmata – where the chromosomes touch
        - ▪ Nuclear envelope breaks down, spindle fibres attach to kinetochores, chromosomes move to central plane in pairs
    - o Metaphase I – paired chromosomes oriented on opposite poles
        - ▪ Terminalisation – chiasmata move towards telomeres
    - o Anaphase I – chromosome disjunction (separation of paired chromosomes), separated homologues move toward opposite poles
    - o Telophase I – chromosomes reach poles, nuclei, spindle disassembled, daughter cells separated, chromosomes decondense, each chromosome has 2 sister chromatids
    - o Prophase II → Metaphase II → Anaphase II → Telophase II → Cytokinesis (same as Mitosis but 1/2)
- Importance – generates genetic variation, increases diversity
    - o Mendelian inheritance: behaviours of Mendel's particles (genes) during production of gametes in peas precisely parallels the behaviour of chromosomes

Mitosis

Diploid (2n) mother cell 2c

DNA synthesis

4c

Division

2c 2c

Identical diploid (2n) daughter cells

Meiosis

2c Diploid (2n) meiocyte

4c

Division I

2c 2c

Division II

c c c c

Nonidentical haploid (n) cells

Central element Lateral elements

Recombination nodule

Transverse fibers

Chromosomal fibers of homolog 1

Chromosomal fibers of homolog 2

(b)

Copyright 2000 John Wiley and Sons. Inc.

Pair of homologous chromosomes

Homologue 1 Homologue 2

Centromeres

One chromatid

Synapsis and crossing over

Chiasma

Two chiasmata

Tetrad

Recombinant chromatids