Lecture 4: Genetics and Diversity

Important Concepts

- DNA is genetic material that is passed between generations (chromosomes)
- Specific DNA sequences (A, G, C, T) form genes that make up an organism
- Genetic diversity arises due to **meiosis** in germ cells (sperm and egg-form haploid gametes
- Individuals inherit two copies of one gene: one from each parent (alleles)

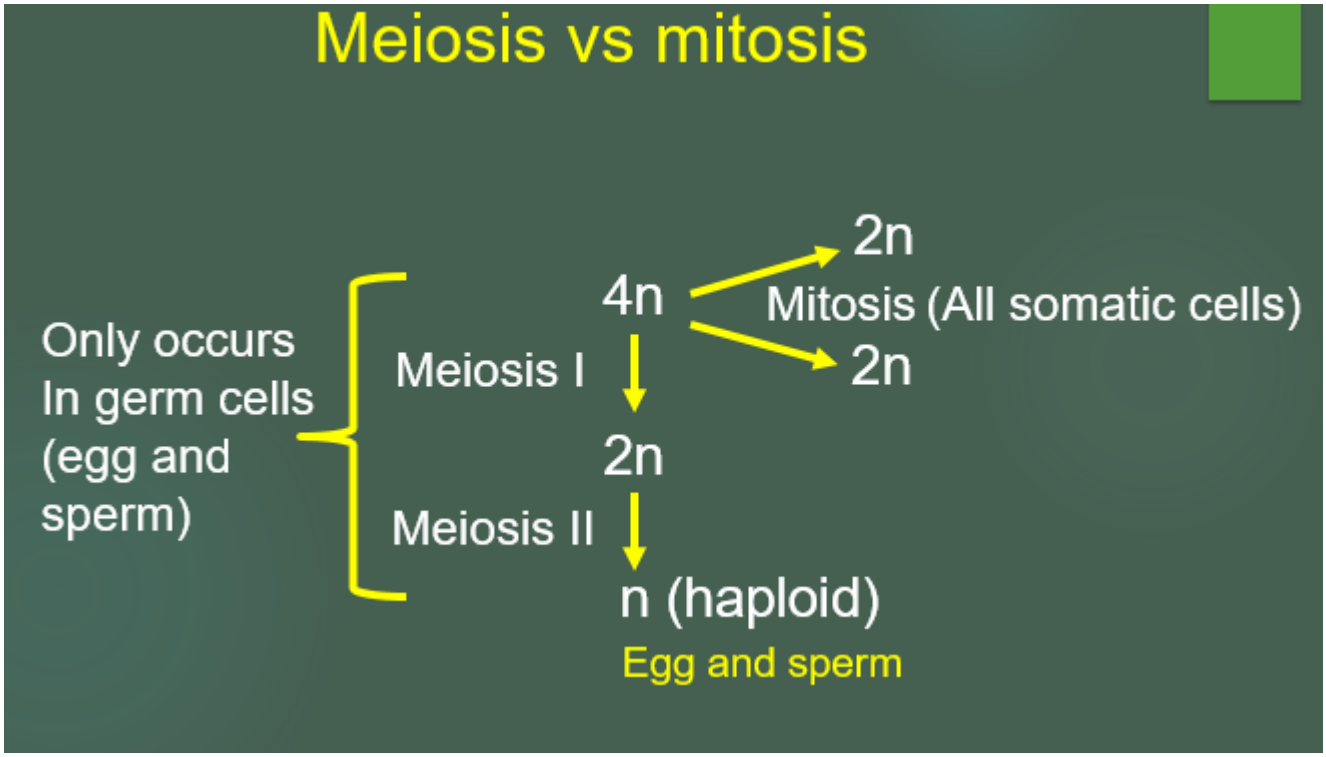
DNA Statistics of Our Genome

- approx. 22 000 genes found on all our 23 chromosomes
- genes are specific sequences of DNA that can be made into proteins (eg hair, eye color, insulin)
- most genes are thousands of bases long, but only account for about 5% of our total genome

Understanding Basic Genetics

- all cells have 2n (diploid) genetic content (2 alleles (versions) of each gene)
- prior to entering mitosis, a cell has 4n genetic content (after S phase)
- to make gametes (egg,sperm), organisms have to go through one additional division in germ cells (ovaries, testes)
- **Meiosis** is process of cell division in germ cells
- during meiosis, chromosomes can exchange DNA (crossing over) which leads to new genetic combinations and diversity

Meiosis vs Mitosis



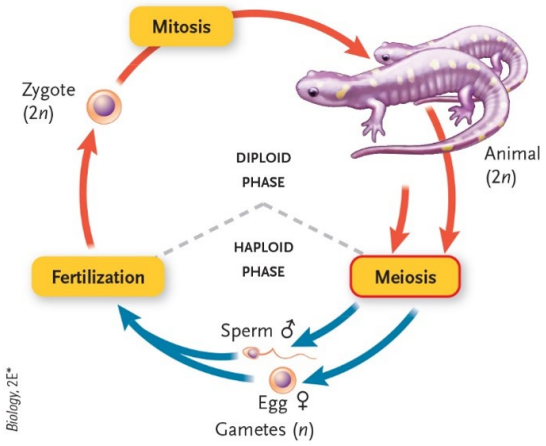


FIGURE 7.4 Animal life cycle. Meiosis results in the formation of haploid gametes that fertilize to form a diploid zygote. Zygotes divide by mitosis. *n* refers to the haploid number of chromosomes; 2*n* refers to the diploid number.

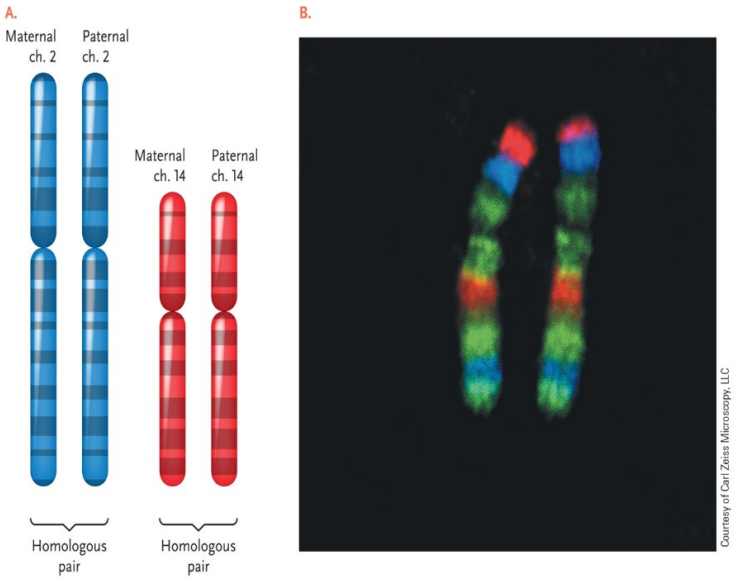


FIGURE 7.5 Homologous chromosomes. **(a)** Schematic diagram illustrating two pairs of homologous chromosomes. Typically, one chromosome of each pair is inherited from the mother and the other from the father. **(b)** Coloured patches in the fluorescence micrograph indicate corresponding DNA sequences on the chromosome. These chromosomes carry the same series of genes, but the DNA sequence of any one of those genes might differ just a bit from that of its partner on the other chromosome. Different forms of a gene are called alleles.

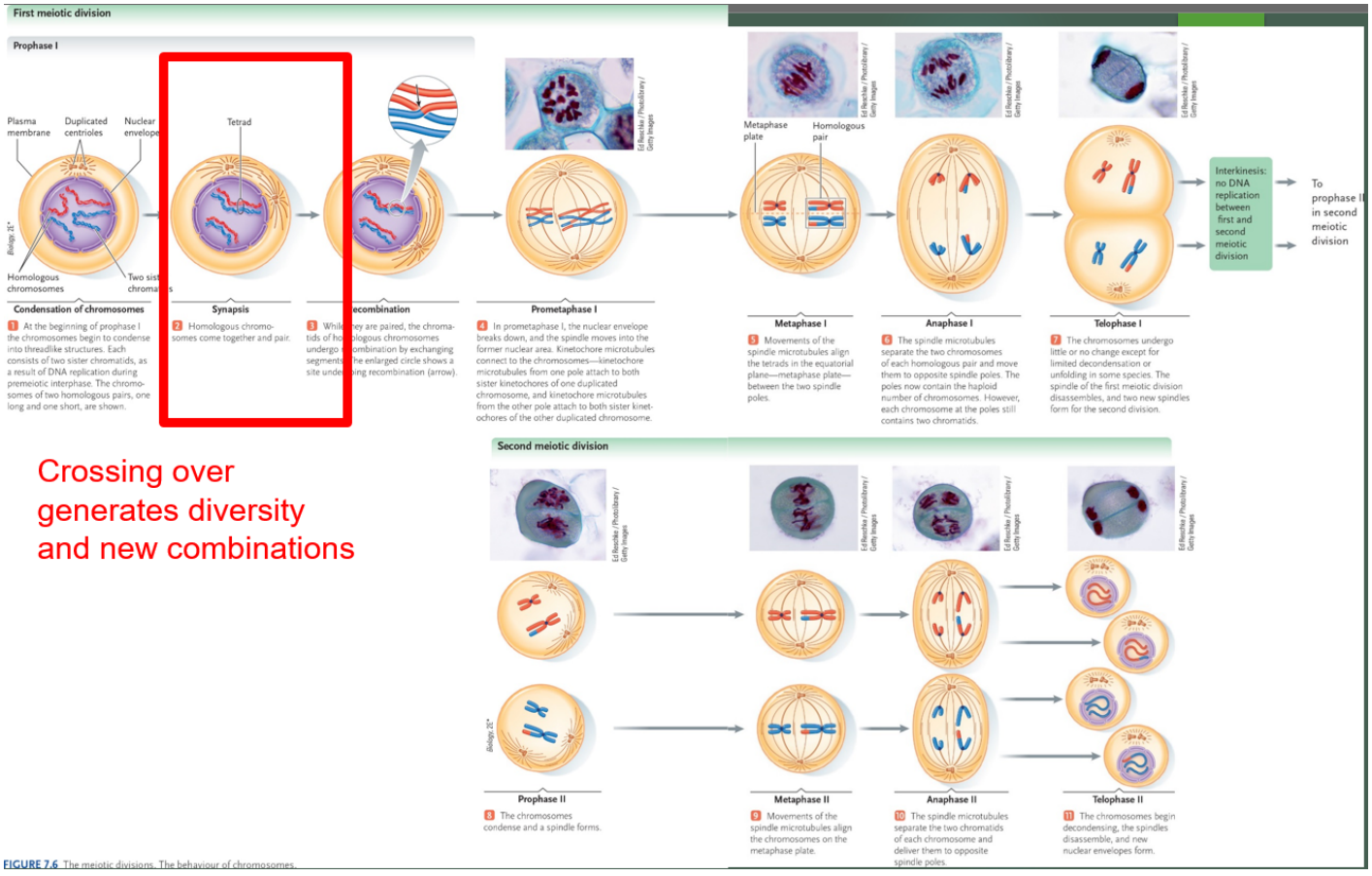


FIGURE 7.6 The meiotic divisions. The behaviour of chromosomes.

Errors During Meiosis

- errors create gametes with an incorrect number of chromosomes (non-disjunction)
- can occur with autosomes (trisomy #21-Downs syndrome)
- can occur with sex chromosomes (XXY-Kleinfelter syndrome)
- **Aneuploidy**: lacking or extra chromosomes
- **Ploypoidy**: extra sets of chromosomes

Genetics & Inheritance

- Gregor Mendel developed genetic laws to describe inheritance of genes
  - used pea plants to describe patterns of specific trait inheritance (eg flower color)
- genes (genotype) control the development of traits (phenotype) $Genes \Rightarrow Phenotype$

Mendel's 4 Genetic Laws

1. **Law of Inheritance**: traits are controlled by genes which are inherited: one from each parent
2. **Law of Dominance**: if two inherited genes are different (alleles), one may be *dominant* over the other one and express this trait
3. **Law of Segregation**: during meiosis, one of each pair of genes will segregate from one another into gametes
4. **Law of Independent Assortment**: one of each pair of genes will segregate independently from one another during meiosis

Sex-linked Diseases

- Two sex chromosomes (X, Y) can alter disease ratios
- X-linked recessive deases (eg hemophilia) are more common in males since only have one X chromosome

Epigenetic Inheritance

- changes in inheritance patters w/o changes in DNA sequence
- due to addition in methyl group (CH_4) onto nucleotide cytosine
- methylation of cytosine affects the turning on and off of genes at specific times and can be influenced by environmental factors
- also may have effects on disease and inheritance