

Genetics Notes Chapter 2

Intro.

- **Gene Discovery**-Finding a subset of genes in a genome that influence a biological property.
- **Single Gene Inheritance**- Genetic conditions caused by a mutation in a single gene follow predictable patterns of inheritance within families.
- **Crosses**-Certain types of controlled matings.
- **Wild type**-An organism with the normal phenotype.
- **Mutant**-Organism with the abnormal phenotype.
- By crossing mutants with wild types and then crossing those offspring one can see if the gene follows a single gene inheritance pattern based on the phenotypic ratios of the progeny.
- First discovered by Mendel, also called **Mendelian Inheritance**.

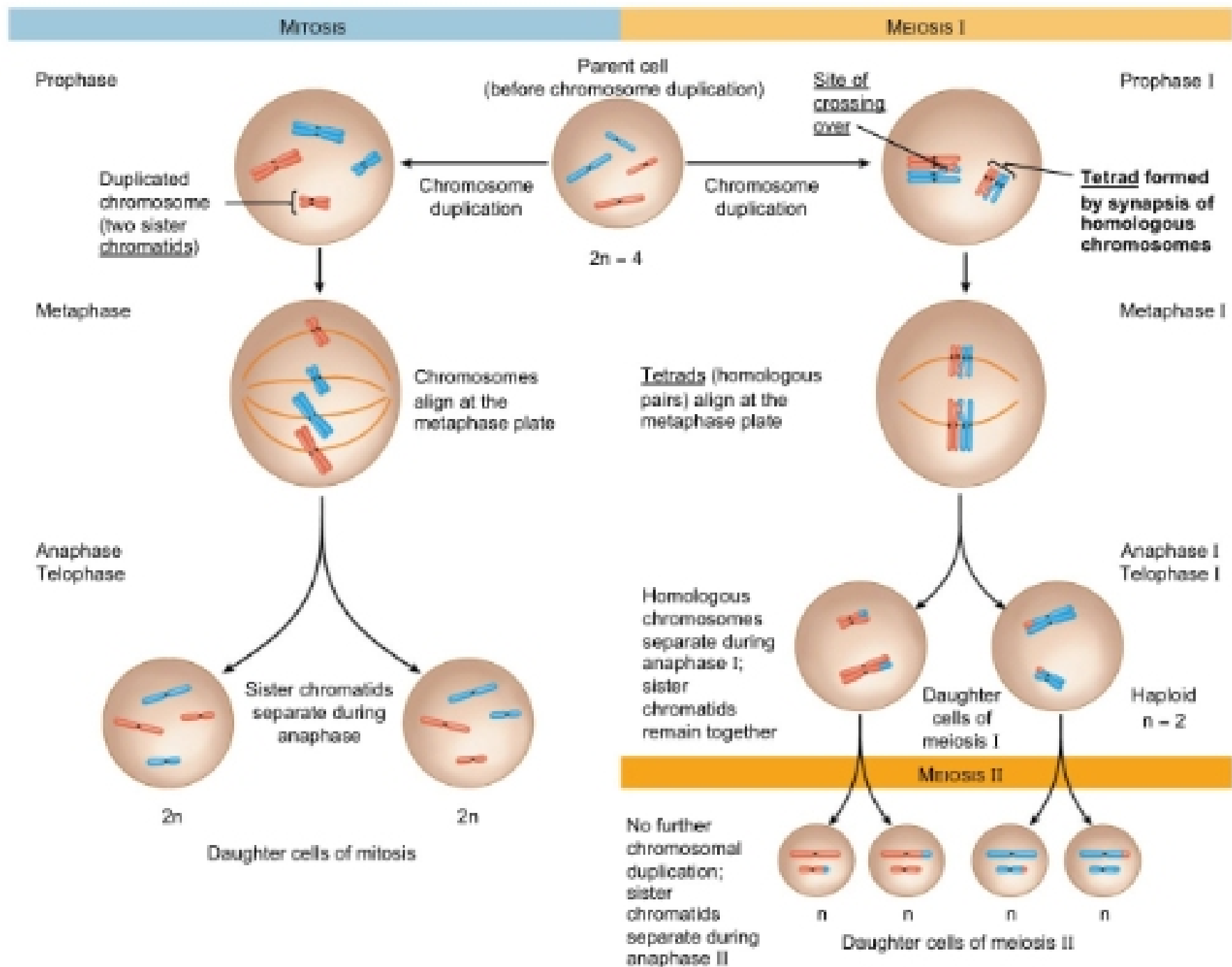
2.1

- Mendel used the **garden pea** in his inheritance experiment.
- He chose to examine seven properties: pea color, pea shape, pod color, pod shape, flower color, plant height, and position of the flowering shoot.
- The terms **character, trait, and property** are used synonymously.
- He studied to contrasting phenotypes of each trait.
- **Phenotype**-the set of observable characteristics of an organism defined by its genotype.
- Mendel used only **Pure Lines**- All progeny produced by matings within members of that line had that trait.
- Two types of pea crosses: **cross pollination**-transfer of pollen from one flower to another.
Selfing-transfer of pollen to fertilize the stigma of the same flower.
- The first cross between two organisms is called the **Parental Generation** (abbreviated P).
- The first generation of progeny is called the **first filial generation** or **F1** and normally all display the dominant phenotype.
- Mendel then crossed individuals from the F1 generation and found that the F2 generation displayed a phenotypic ratio of very close to $\frac{3}{4}$ to $\frac{1}{4}$.
- **Mendel's Laws of Equal Segregation**
 - 1. A gene is necessary for producing a phenotype (like gene color)
 - 2. Each plant has a pair of this type of gene.
 - 3. The gene comes in two forms (**alleles**) and can be represented by a "Y" gene and a "y" gene.
 - 4. A plant can be either Y/Y, Y/y, or y/y.
 - 5. The "Y" allele is **dominant** while the "y" allele is **recessive**.
 - 6. In meiosis the members of a gene pair separate equally into the eggs and sperm (known as **Mendel's law of equal segregation**).
 - 7. A single gamete contains only one allele.
 - 8. At fertilization gametes randomly fuse regardless of which alleles they bare.
- **Zygote**-a fertilized egg.
- **Homozygote**- An organisms with a pair of the same alleles.
- **Heterozygote**- Organisms with a pair of two different alleles.
- Can be homozygous dominant Y/Y, heterozygous Y/y, or homozygous recessive y/y.
- The allelic combinations that underlie phenotypes are called **genotypes**.

- A cross between the type Y/y and Y/y is known as a **monohybrid cross**.

2.2

- Gene pairs are located on chromosome pairs. It is the **chromosome pairs** that segregate, carrying the genes with them during meiosis.
- When somatic cells divide they undergo mitosis.
- Mitosis can occur in both haploid and diploid cells resulting in 2 daughter haploid or diploid cells.
- Most eukaryotes also have specialized diploid cells called **meiocytes** which produce sex cells such as sperm, eggs, or fungal spores.
- Two sequential cell divisions occur in a process called meiosis. $2n \rightarrow n+n+n+n$



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- Replicate sister chromosomes are called a **dyad**.
- In meiosis during metaphase 1 two homologous dyads pair up and form a **tetrad**.
- Final result is four haploid daughter cells each with one of the chromatids from the tetrad.
- Spindle fibers that separate the chromatids during anaphase are made up of tubulin. The fibers are depolymerized (shortened) at the site where they are attached to the chromosome.

- Haploid organisms serve as good model organisms because there is no consideration as to male or female sex. Additionally the genotype is not masked in the phenotype by any dominance.

2.3

- Alleles at the molecular level are sections of genes. The mutant and wild-type alleles differ by only a few nucleotides of sequence out of the several thousands of nucleotides that make up the gene.
- Changes in a nucleotide sequence that result in a new allele are called mutations.
- Mutations can occur along multiple different places in the allele's DNA sequence.
- Alleles with mutations are normally recessive because it usually only takes one wild-type allele to provide normal function.
- Replication of DNA occurs during the "S" phase.
- Most mutations alter the amino acid sequence of the gene's protein product resulting in **null (zero) function** or **leaky (some) function**.
- Recessiveness of a mutated allele is observed in heterozygote homologs that are **haplosufficient**. This means that the single wild-type allele provides enough product (normally a protein) to provide normal function.
- Dominance of a mutated allele is observed in heterozygote homologs are **haploinsufficient** because the one wild-type allele does not provide for the creation of enough product for normal function.

2.4

- Forward genetics:
- Choose phenotype of interest
- Find mutants affecting that property
- Check and do crosses to see if it follows single gene inheritance patterns
- Identify time and place of action for genes
- Do DNA analysis
- **Testcross**-Crossing one parent with the dominant phenotype (but unknown genotype) with a recessive. If the progeny are 1:1 phenotype the parent is heterozygous if the progeny shows all dominant phenotype then the parent is homozygous dominant.
- If a recessive tester is unavailable the organisms of dominant phenotype but unknown genotype can be selfed if the progeny produced is 3:1 then it is a heterozygote.
- The principles of inheritance can be applied in two directions:
- Inferring genotypes from phenotypic ratios
- And predicting phenotypic ratios from parents of known genotypes.

2.5

- Most plants and animals show **sexual dimorphism** (male and female)
- Females have 2 X chromosomes, males have an X and a Y chromosome.
- Females are the **homogametic sex** because they only produce gametes with X chromosomes.
- Males are the **heterogametic sex** because they produce 50% gametes with the X chromosome and 50% with the Y chromosome.