Genetics Notes Chapter 17: Large-Scale Chromosomal Changes

Introduction

- Chromosomal mutations are changes in a **chromosome region encompassing multiple genes**.
- In contrast, gene mutations take place within a single gene.
- Many chromosome mutations are detectable with microscopy.
- Two types of chromosome mutations:
    - Changes in **chromosome number**
    - Changes in **chromosome structure**

14.1 Changes in Chromosome Number

- **Euploid**-Organisms with multiple sets of their genome. (haploid, diploid etc.)
- **Aberrant Euploids**- Have more or fewer than the normal sets of chromosomes.
- **Polyploids**- Have more than two sets of chromosomes (triploid, tetraploid, pentaploid)
- **Monoploid**-An organism that is normally diploid, but abnormally has only one set of chromosomes (different from haploid).
- Male bees, wasps, and ants are monoploid, they are produced through **parthenogenesis**. The unfertilized egg developing into an embryo.
- Normally monoploids do not develop, or develop many abnormalities because they have many deleterious mutant alleles that are not masked by a second set of genes. Monoploids are normally sterile because their chromosomes don't have pairing partners for meiosis. Male bees bypass this is produced through mitosis.
- Polyploidy is very common in plants. It often causes new species of plants to arise.
- The number of genome sets correlate with the size of the organism.
- **Autoplyploids**-Have multiple chromosome sets originating within one species.
- **Allopolyploids**-Have multiple chromosome sets from two or more closely related species. These chromosomes are only **homeologous** (partly homologous).
- In a triploid there are **trivalents** or paired groups of three. These separate randomly in meiosis so that some gametes get two alleles of the gene and the other gets one. Overall both gametes have intermediate size between the haploid and diploid size. These are **aneuploidy**.
- **Aneuploids** typically do not produce viable offspring.
- Autotetraploids can occur spontaneously but can also be induced through a chemical that blocks the spindle fibers from working.
- When spindle fibers are blocked the chromosomes cannot properly segregate but mitosis still continues forming a tetraploid.
- One such chemical that blocks the formation of spindle fibers is called **colchicine**.
- If during meiosis the tetraploid chromosomes pair as two bivalents then 2n gametes are produced.
- If the tetraploid chromosomes pair as a quadrivalent then there are also 2n gametes.
- If the tetraploid pairs as a univalent and a trivalent then aneuploid gametes are created that are nonfunctional.
- An allopolyploid is a hybrid plant of two or more species containing two or more copies of each of the input genomes.
- **Amphidiploid**-A doubled diploid $2n_1+2n_2$ gametes are n_1+n_2

- 50% of angiosperms are polyploidy. The larger genomes lead to greater variation and perhaps greater adaptability?
- Allopolyploids can be created by crossing related species to double the chromosomes of the hybrid or by fusing diploid cells together.
- Monoploids can be cultured by subjecting haploid pollen grains to cold temperatures causing them to multiply and form an embryoid. This embyoid can be grown to form a monoploid plantlet.
- These monoploids are used to look for desirable allelic combinations and to see if favorable mutations can be induced.
- If these plants have favorable traits their chromosomes can be doubled using colchicine and used to grow diploid crops.
- Seedless fruits are normally sterile triploid organisms.
- Some animals are also naturally and artificially polyploid.
- A aneuploidy is an organism that differs from the wildtype chromosome number by part of a chromosomal set.
  - 2n+1 is **trisomic**
  - 2n-1 is **monosomic**
  - 2n-2 (-2 represents loss of both homologs) is **nullisomic.**
  - In haploids n+1 is **disomic.**
  - Mutations of sex chromosomes include: XXY, XYY, XXX, or XO (O stands for sex chromosome absence).
- A common cause of aneuploidy is **nondisjunction**, or the failure of two chromosomes or sister chromatids to segregate resulting in two going to one pole and none going to the other.
- Mitotic nondisjunction results in sections of aneuploid cells called aneuploidy sectors.
- Meiotic nondisjunction is more common producing n-1 gametes and n+1 gametes. When fertilized with normal gametes they produce 2n-1 (monosomic) zygote or 2n+1 (trisomic) zygote.
- Crossovers are needed to keep bivalents paired until anaphase 1 in meiosis. If crossing over fails there is no chiasmata formed which can lead to first-division nondisjunction the most common type.
- In monosomics (2n-1) the lack of one chromosome is normally lethal or deleterious.
- Monosomics of the X chromosome can survive, and have **Turner's syndrome.** Represented as XO these individuals are sterile, and have many characteristic features.
- Trisomics (2n+1) can be both lethal and survivable. Includes **downs syndrome**, **Klinefelters syndrome** XXY.
- Aberrant euploids (in plants) tend to have larger size but similar proportions and shape. However, aneuploids in plants An anueploid plant has different proportions from the wildtype.
- In animals, trisomics are usually less lethal than monosomics.
- Euploids have **gene balance.** Any number of genes on and two chromosomes are 1:1 no matter if diploid or triploid or tetraploid etc.
- Aneuploid genes are out of balance.
- Some genes (that are more important) contribute more than others to this gene imbalance.
- These genes are **haplo-abnormal** if they result in an abnormal phenotype when they only appear once and some genes are **triplo-abnormal** if the abnormal phenotype is the result of three copies of the gene.

17.2-Changes in Chromosome Structure

- Chromosome segments can be:
  - Lost-**Deletion**
  - Doubled-**Duplication**
  - Reversed-**Inversions**
  - Moved to a different chromosome-**Translocation**
- These chromosomal rearrangements are caused by a **double stranded break** in the DNA and then rejoining of this segment.
- These chromosome rearrangements normally only survive if the chromosome retains its singular centromere and two telomeres.
- If the chromosome is **acentric** (it lacks a centromere) it will not be pulled to either pole during meiosis.
- If it is **dicentric** (has two centromeres) meiosis cannot properly occur.
- Without telomeres the DNA cannot properly replicate.
- These rearrangements may also cause gene imbalance and therefore abnormalities.
- Another rearrangement is caused by crossing over between duplicated sequences. The chromosomes may align with similar sequences that are located at different positions on the chromosomes. This is nonallelic homologous recombination.
- Rearrangements can be unbalanced and balanced:
  - **Unbalanced** causes a difference in gene dosage. These classes are gene duplication and gene deletion.
  - **Balanced** rearrangements change the chromosomal gene order but do not remove or duplicate DNA. These are inversions and reciprocal translocations.
- A small deletion within the gene is called an **intragenic deletion** and is able to inactivate the gene.
- **Multigenic deletions** are deletions of large segments of DNA. They are almost always lethal in the homozygous and often lethal or deleterious in the heterozygous due to gene dosage imbalance.
- If you have 2 homologs, one with dominant wildtype, and the other with recessive mutations normally the dominant wildtype phenotype will be displayed. But if two of the mutant phenotypes are shown then this is a clue that this section on the dominant wildtype chromosome may have been deleted, an effect termed **pseudodominance**.
- These deletion mutations often arise spontaneously in the sperm or eggs of the parents.
- Duplication rearrangements can be either right next to the original segment (**tandem duplications**) or inserted elsewhere into the genome (**insertional duplication**).
- **Duplication and deletion loops** arise in meiosis when a section of the DNA is looped out because there is no homologous region on the second chromosome.
- Segmental duplications are common in humans and consist of large duplications anywhere from 10 to 50 kilobases in length that can be seen as both tandem and insertional sequences.
- In an inversion a DNA segment is cut out, flipped, and reinserted.
  - If the inversion is outside the centromere it is **paracentric**.
  - If it spans the centromere it is **pericentric**.