PCB 3063: Exam Two Study Guide

Sex Determination & Sex Chromosomes (Chapter 5)

- **Know the difference between autosomes and sex chromosomes, understand the terms heterogametic sex and homogametic sex, and know the difference between XY and ZW sex determination systems**
  - **Autosomes**: Any chromosome not considered a sex chromosome
  - **Sex Chromosomes**: chromosomes (e.g. X & Y) that are involved in sex determination
  - **Heterogametic Sex:** sex of a species possessing dissimilar sex chromosomes
    - E.g. Males: **XY**
  - **Homogametic Sex:** sex of a species possessing similar chromosomes
    - E.g. Females: **XX**
  - The **XY sex determination system** is more commonly known to occur in mammals and some insects
    - **XX** determines a female; **XY** determines a male
  - The **ZW sex determination system** commonly applies to reptiles, birds, & some insects and amphibians
    - **ZW** determines a female; **ZZ** determines a male
  - **Key Point:** In the XY system, females are **homogametic**, but in the ZW system, females are **heterogametic** and vice versa for males

- **Know the evidence which indicates that the Y-chromosome determines sex in mammals (existence of Turner and Klinefelter individuals), know what the genetic makeup is of people with Turner and Klinefelter Syndromes and what the characteristics (phenotypes) of these syndromes are**
  - **Absence of the Y chromosome** results in gonads forming into ovaries and completion of female development
  - **Presence of the Y chromosome** results in gonads forming into testes and completing male development
  - **Turner Syndrome: X**
    - Short stature, webbed neck, rudimentary ovaries, underdeveloped breasts
  - **Klinefelter Syndrome: XXY**
    - Tall stature, minor breast development, testicular atrophy, female pubic hair pattern

- **Know the difference in the genes that are carried by the X and the Y chromosomes, know what the PAR region is on the Y chromosome and why it is important.**
  - X chromosomes carries thousands of genes but no genes coding for sex determination, whereas the Y chromosome only has about a several dozen active genes and is much smaller compared to an X chromosome
  - The Y chromosome contains the **SRY gene**, which is responsible for male development
  - Both the X & Y chromosomes do contain a small region in which there is gene homology, which allows the X and Y chromosome to pair and properly segregate during meiosis in males

        - This small region is known as **PAR (pseudo autosomal region)**

- **Understand how the SRY gene controls sex determination in mammals (initiates male development through regulation of testosterone and antimullerian hormone production during early development) and what the evidence is for the importance of this gene (XY females and XX males). Know where the SRY gene is located on the Y chromosome.**

    - The **SRY gene** codes for a protein, a transcription factor, that regulates the transcription of genes that are needed for male hormone development
    - Cells in testis produce and release **anti-Mullerian hormone**, which blocks Mullerian ducts from developing into uterus and ovaries, arresting overall female development
        - **Testosterone** is also secreted, and stimulates the formation of internal and external male structures
    - The SRY gene (or testis-determining factor) is near the boundary of the **PAR**
    - The **close proximity** of the SRY gene and PAR can result in XX males and XY females when crossover occurs between an X and a Y chromosome
        - XX males have a translocation from the Y to the X
        - XY females have deletion of part of the Y
    - **Androgen Insensitivity** – Testicular Feminization
        - Mutation in an autosomal gene coding for the testosterone receptor
        - Karyotype is XY; however, externally appears female & contain underdeveloped testis in abdominal cavity

- **Understand the concept of dosage compensation and why it is important on the X chromosome and how dosage compensation is achieved in mammals (X-inactivation) and in fruit flies (increase of activity of genes on the X in males).**

    - **Dosage compensation** refers to the equalization of gene expression between the males and females of a species
    - Because sex chromosomes contain different numbers of genes, different species of organisms have developed different mechanisms to deal with this inequality
    - For mammals who have the XY sex determining system, the homogametic species (females, XX) inactivate one of their X chromosomes in order to achieve equal gene expression as their heterogametic counterparts, the XY males, whom only possess one X chromosome to begin with
    - Fruit flies (Drosophila), who also have the XY sex determining system, go about neutralizing the gene expression between males and females in a different manner
        - Since an XY karyotype in drosophila still represents a male, the activity of genes on that single X chromosome is doubled to compensate for a lack of an X chromosome

- **Know what a Barr body is and how inactivation of the X chromosome occurs during early development in mammalian females, and be able to state how many Barr bodies would be present in a cell that has 1, 2, 3, or 4 X chromosomes. Understand the term genetic chimera (genetic mosaic), and be able to explain the pattern of inheritance of calico and tortoiseshell**

coat colors in cats

- **Barr Body:** the inactivated X chromosome in a female (XX) during early development
- Number of Barr Bodies = Number of total X chromosomes **minus 1**
  - Only **ONE** X chromosome is left active & functional
- X-inactivation occurs during the late blastula stage of development
  - Both X chromosomes are turned on, but **ONE** X is turned off
  - The X chromosome that is turned off in any given cell is random choice
- The pattern of inheritance of calico in female cats can be attributed to the random X inactivation of the coat color genes which is X-linked
  - Because the X that is inactivated is random, female cats will display a random pattern of orange and black fur depending which allele is expressed on each part of the body
  - The same holds true for tortoiseshell coat color genes
- Female cats are known as **genetic mosaics** (**chimeras**) because of this random display X-linked coat color genes across their body
  - Human females are also genetic mosaics (e.g. Ectodermal dysplasia)

➢ **Know what the XIST gene does and where it is located**

- X-inactivation begins at the **XIC (X-inactivation Center)**
  - Near the centromere on the X chromosome
- **XIST (X-inactivation Specific Transcript):** gene responsible for X-inactivation
- During X-inactivation, on one of the two X chromosomes, XIST turns on
- When the XIST gene is turned on, the chromosome it sits in shuts down
  - XIST **is transcribed** to RNA, but **not translated** to protein
  - The DNA of the inactive X becomes methylated

➢ **Be able to provide an explanation of why individuals with Turner and Klinefelter syndromes are not phenotypically normal.**

- Not all genes on the inactive X are turned off
- Individuals with Turner and Klinefelter experience either more or less gene expression depending how many extra X's are present or absent

Chromosome Mutations (Chapter 6)

➢ **Know the following terms and their significance: Chromosome mutations or chromosome aberrations, Nondisjunction,Aneuploidy, monosomy, trisomy,Down's Syndrome, trisomy 21, Amniocentesis,Triploid, tetraploid, and polyploid**
Chromosome deletion, duplication, inversions, translocations Familial Downs Syndrome

- **Chromosome aberration (mutation):** a chromosome missing extra, or irregular portion of chromosomal DNA
  - It can be due to an atypical number of chromosomes or a structural abnormality