Inheritance Patterns & Human Genetics
NOTES

Mutations may occur in a:

1. **Germ cell – Occurs in a gamete (egg or sperm): passed on to offspring.**
2. **Somatic cell – Occurs in a body cell: can not be passed on to offspring.**

Mutations may cause:

1. **Death**
2. **Not Harmful**
3. **Harmful (disease or disorder)**
4. **Beneficial (few) (improve chances of survival)**

Mutation Types:

1. **<u>Cell mutation</u> - Occurs in body cells of germ cells (The actual cell is changed)**
    a. **Germ cell – (egg or sperm): passed on to offspring.**
    b. **Somatic cell – body cell: can not be passed on to offspring.**

2. **<u>Chromosome mutation</u> – Changes in the structure of a chromosome or loss of a chromosome**
    a. **Deletion – a portion of the DNA is deleted**
    b. **Inversion – a portion of the DNA coded is reversed**
    c. **Translocation – a portion of the DNA is taken out and transferred to a new location**
        i. **deletion, inversion & translocation mutations change the codons of DNA possibly coding for a different protein.**
    d. **Nondisjunction – abnormal segregation of chromosomes during meiosis. This will cause an extra chromosome or one less chromosome in a gamete (sex cell).**

3. **<u>Gene (DNA) mutation</u> – May involve segments of DNA or a single nucleotide of DNA.**
    a. **Point mutation – change in the DNA code at one point in the sequence**
        i. **Example – Sickle cell anemia**
    b. **Frame shift mutation – deletion or addition of a nucleotide in DNA and this changes the codons. (Changes the protein!)**

<u>Human Genetics:</u>

Studying Human Inheritance – Humans have 25,000 to 30,000 genes.

- **Pedigree – a family record that shows how a trait is inherited over several generations.**
- **Carriers – individuals who have one copy of a recessive allele. This individual may carry the gene for the disorder but does not show the trait but may pass the trait to their offspring.**

Types of Genetic Disorders: **(Some are normal traits not disorders)**

1. **Dominant Allele – caused by inheriting a dominant gene.**
    a. **Huntington's disease – There is a genetic marker (section of DNA) that can identify the presence of Huntington's disease.**
2. **Recessive Allele – caused by inheriting two recessive genes for the disease.**
    a. **Cystic Fibrosis**
3. **Codominant Allele – caused by inheriting two dominant genes for the trait.**
    a. **Sickle Cell Anemia**
4. **Multiple Allele – caused by inheriting two of the three or more alleles for the trait.**
    a. **Human Blood Type (Not a disorder)**
5. **Polygenic traits – controlled by two or more genes.**
    a. **Skin color, Eye color, Height**
        i. **Melanin – pigment in skin and eye color**
6. **X-linked Trait – caused by inheriting a recessive gene on the X chromosome.**
    a. **Color Blindness, Hemophilia, Duchenne Muscular Dystrophy, Lesch-Nyhan disease**
7. **Sex Influenced Trait – caused by inheriting a set of alleles that are influenced by male or female sex hormones.**
    a. **Pattern Baldness**
8. **Nondisjunction – caused by abnormal segregation of chromosomes during meiosis. This will cause an extra chromosome or one less chromosome in a gamete (sex cell).**
    a. **Monosomy – 45 chromosomes, the person is lacking one chromosome**
    b. **Trisomy – 47 chromosomes, the person has an extra chromosome**
        i. **Down's Syndrome – Trisomy 21**
        ii. **Klinefelter Syndrome – XXY**
        iii. **Turner's Syndrome – XO**
        iv. **OY – Do not survive**

Detecting Genetic Disorders:

Step One: Genetic Screening: take a family history and progress to genetic tests

- **Tests:**
    - **Amniocentesis**
    - **Chorionic Villi Sampling**
    - **Blood Sampling: check for protein markers**

Example – Newborns are screened for PKU (phenylketonuria). PKU is a child's inability to metabolize phenylalanine found in milk and NutraSweet. They have to be placed on a diet without these foods or severe mental retardation will occur.

Step Two: Genetic Counseling: medical guidance that informs them about problems that could affect their child.