N201 – Special Populations
Heredity

I. Definition
- A. Genetic disorders/ inherited disorders are disorders that can be passed from one generation to the next
  1. Disorder in the gene or chromosome structure
  2. Alterations of a whole chromosome, a part of a chromosome or even a single gene can manifest as a genetic disorder
- B. Genetics: the study of the way such disorders occur

II. Basic genetics
- A. Genes
  1. Basic units of heredity
  2. Determine physical & mental characteristics
- B. Chromosomes
  1. 44 autosomes (22 pair)
  2. 2 sex chromosomes (1 pair)
     =46 total chromosomes for humans
  3. Determining sex of the infant:
     a. if sex chromosomes are both type XX in the zygote formed from the union of sperm and ova then the infant will be female
     b. if one is X and one is Y then the infant is male
- C. Phenotype- outward appearance or the expression of genes
- D. Genotype- actual genetic composition or makeup of an organism
- E. Genome- complete set of genes present in an organism (complete set DNA)
- F. Medelian Inheritance
  1. Principles of genetic inheritance of disease are the same as these that govern genetic inheritance of other physical characteristics
  2. <u>Homozygous</u>
     a. two like genes for a trait
  3. <u>Heterozygous</u>
     a. unlike genes for a trait
  4. <u>Dominant</u>
     a. dominant in their action over others
  5. <u>Recessive</u>
     a. a gene that is not dominant

III. Inheritance patterns
- A. Example One
  1. Father is homozygous dominant
  2. Mother is homozygous recessive
  3. Prediction

IV. Autosomal dominance
- A. Many disorders are known but only a few commonly seen
- B. Person with dominant gene for a disease is usually heterozygous
  1. Huntington's chorea
  2. Osteogenesis Imperfecta
  3. Marfan syndrome

- C. Example One:
  1. Father dominant disorder
  2. Mom does not have a trait
  3. Prediction:

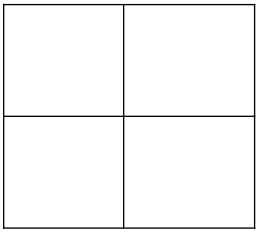
- D. Example Two
  1. Mom dominant inherited disorder
  2. Father dominant inherited disorder
  3. Prediction

- E. Dominance inherited pattern is present
  1. Sex of the individual is unimportant
  2. Usually a history of disorder in other family members

V. Autosomal Recessive
- A. Most genetic disorders are inherited as recessive traits vs dominant traits
- B. Does not occur unless 2 genes for the disease are present

    1. Many inborn errors of metabolism
        a. Cystic fibrosis
        b. Tay sachs
        c. Galactosemia
        d. PKU

- C. Example One
    1. Both parents heterozygous in genotype
    2. Both parents disease free
    3. Both carry a recessive gene for the disease
    4. Prediction



- D. Example Two
    1. Mother disease free but recessive gene for disease
    2. Father no traits
    3. Prediction

VI. X- linked Dominant (sex linked dominant)
- A. Some genes for disorders are located on and transmitted only be female sex chromosome (X)
- B. Ex: hypophosphatemia

VII. X-linked recessive
- A. Majority of X-linked disorders are recessive
- B. Female - normal gene also present blocks expression of the disease
- C. Only male children will have the disease
    1. Hemophilia
    2. Muscular dystrophy

VIII. Multifactoral (Polygenic)
- A. Congenital disorders present at birth:
    Heart disease, HTN, DM
    Pyloric stenosis, cleft lip/palate
    Neural tube defects (NTD)
    Mental illness

B. Environmental influences determine if the disorder is expressed

IX. Chromosomal Abnormalities
A. In some cases of genetic disease the abnormality occurs not because of dominant or recessive gene patterns, but through a fault in the number or structure of chromosomes.
1. Extra chromosomes-
2. Missing chromosome pieces-

X. Diagnostic Testing
A. Karotyping
1. Visual presentation of a person's chromosome pattern
2. Blood sample or scraping of cells from buccal membrane, stain, magnify, photograph

B. Barr body determination
1. Determine the sex of a child
2. Scrape cells from buccal cavity, stain, magnify

C. Alpha-Fetoprotein Analysis (AFP)
1. Glycoprotein produced by fetal liver is present in amniotic fluid or maternal serum
2. Screening test time frame:
3. If abnormal: _____

D. Amniocentesis
1. Time frame: 15-20 weeks gestation
2. Locate pocket of amniotic fluid via ultrasound
3. Risks-
4. Post procedure-

E. Chorionic Villi Sampling
1. Retrieve and analyze chorionic villi for chromosome analysis. Ideally done between 10-13 wks gestation.
2. Risks-
3. Post procedure-

F. Cell Free Fetal DNA Screen
1. Blood test- screening method to look for chromosomal abnormalities

XI. Genetic counseling
A. Purpose-to help make accurate decisions about future reproduction
B. Timing- Ideal time is before the first pregnancy
C. Nursing Considerations
1. Support
2. Teaching
3. Do not impose own values and opinions on others