**N201**
**Heredity- 2024**

I. Overview
   A. Genetic disorders cross the mind of pre-pregnancy women and pregnant women
   B. Many advances in genetic screening have been made
   C. What is genetics?
   D. What are genes?

II. Basic genetics
   A. 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

   B. Phenotype- outward appearance or the expression of genes

   C. Genotype- actual genetic composition or makeup of an organism

   D. Genome- complete set of genes present in an organism (complete set DNA)

   E. Medelian Inheritance: Ability to predict inheritance.
      1. <u>Homozygous</u>
         a. two like genes for a trait

      2. <u>Heterozygous</u>
         a. unlike genes for a trait

      3. <u>Dominant</u>
         a. dominant in their action over others

      4. <u>Recessive</u>
         a. a gene that is not dominant

III. Inheritance patterns
   A. Example One
      1. Father is homozygous dominant  (B) Brown eyes
      2. Mother is homozygous recessive  (b) Blue eyes
      3. Prediction

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IV. Autosomal dominance

A. Dominant gene = causes disease expression

B. One of the parents will have the disease

C. Examples:

1. Huntington’s chorea
2. Osteogenesis Imperfecta
3. Dwarfism

- Usually a family history of the disorder
- Both sexes affected

D. Example One: D= Disease, h= Healthy

1. Father dominant disorder (Dh)
2. Mom does not have a trait (hh)
3. Prediction:

E. Example Two

1. Mom dominant inherited disorder (Dh)
2. Father dominant inherited disorder (Dh)
3. Prediction

V. Autosomal Recessive

A. Does not occur unless 2 genes for the disease are present

B. Heterozygous people are unaffected – they carry the trait

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 (Hd)
2. Both parents disease free
3. Both carry a recessive gene for the disease
4. Prediction

D. Example Two
  1. Mother disease free (Hd) but recessive gene for disease
  2. Father no traits (HH)
  3. Prediction

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VI. X- linked Dominant (sex linked dominant)
  A. Transmitted by female sex chromosome
  B. Gene presence on one of the X chromosome = symptoms
  C. Caused by variants in genes on the X chromosome
  D. Dominant, so affected parent will have disease
  E. Examples:
    1. Vitamin D- resistant rickets and Rhett syndrome

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. Fragile X syndrome

VIII. Chromosomal Abnormalities
  A. Numerical OR structural
  B. Abnormal formation of chromosomes
  C. Causes profound abnormalities/ disabilities

IX. 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, and photograph

  B. Alpha-Fetoprotein Analysis (AFP)
    1. AFP is a glycoprotein produced by the fetal liver
    2. Present in the amniotic fluid or maternal serum
    3. Tested 15-20 weeks (16-18 ideal)
    4. Serum test most common
    5. If increased- possible NDT
    6. False positives occur especially if date of conception is not accurate

  D. Amniocentesis
    1. Done After 15 weeks
    2. Placenta location via U/S
    3. Can detect NTD's
    4. Risks- leakage of amniotic fluid, bleeding, infection, placental abruption, miscarriage

5. Post procedure – Monitor FHR, Rhogam if RH-, light activities x 24hr to ↓ uterine irritability

E. Chorionic Villi Sampling
1. 10-13 weeks gestation
2. Chromosomal analysis to look for defects
    a. Testing the chorionic villi from the placental tissue
3. Disadvantage – <u>cannot</u> detect Neural Tube Defects (NTD)
4. U/S for placental location & uterine position
5. Post procedure –RhoGam if Rh-negative

F. Cell Free Fetal DNA Screen
1) A simple blood sample is taken from the mother that is tested for fetal genetic defects by testing fetal DNA that has mixed with the mother's blood
    a) Aneuploidy- extra or missing chromosomes
2) 11-13 weeks gestation
3) Tests for:
    a) Sex
    b) Fetal Rh status
    c) Paternally transmitted single gene disorders
4) Screening test only
    a) If positive- amniocentesis or CVS

X. Genetic counseling
1. Purpose
    a. Estimation of risk for producing a child with a disease
2. Timing
    a. Before conception
3. Nursing Implications
    a. Nurses need awareness of recent advances in genetic testing
    b. Support & Understanding
    c. Teaching