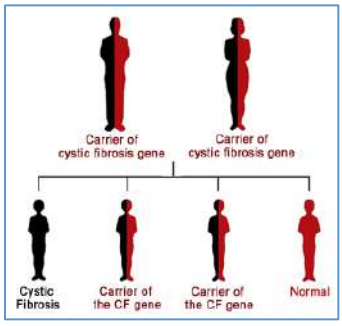
Name _____ Date _____ Block _____

Chapter 11 – Human Genetic Disorders – CP Biology

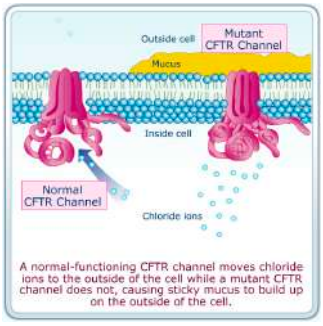
1. Major types of genetic disorders:
   - _____
     - _____
     - _____
   - _____
   - _____

2. Autosomal genetic disorders are caused by _____
   - Most are _____ (need 2 recessive alleles to have the disorder)
     - People with 1 recessive allele are _____ - they do NOT have the disorder but are able to _____
     - Ex: cystic fibrosis (CF), sickle-cell anemia
   - Can also be _____ (need only 1 allele to have the disorder)
     - Ex: Huntington's disease

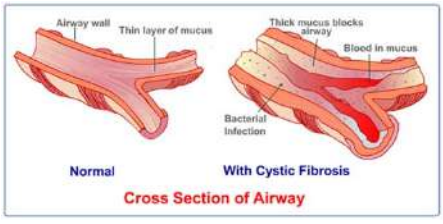
A) Cystic Fibrosis

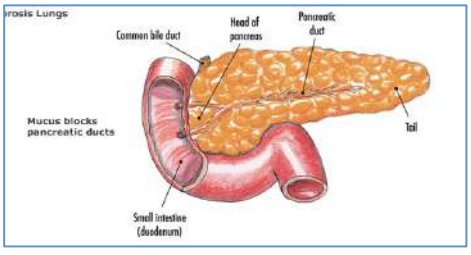


- CF is the most common genetic disorder among _____
  - ~1 in 2500 white infants in the US are born with CF (4-5 born each day)
  - It is estimated that 1 in 20 white people is a carrier of the CF allele
- Caused by an abnormal gene on _____
  - The gene is for a _____ that uses _____ _____ to regulate the movement of sodium (Na^+) and chloride (Cl^-) ions into and out of cells
  - In healthy individuals, the normal protein _____ _____
    - Keeps mucus thin and _____
  - With CF, not enough Cl^- ions are pumped out of cells
    - _____ in airways & pancreatic ducts



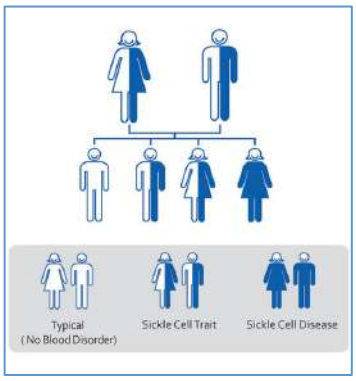
- Symptoms of CF:
  - Buildup of mucus in _____
    - Difficulty _____
    - _____
  - Blocks _____ (produced by the pancreas) from entering the intestine
    - _____
  - Abnormal Na^+ transport also results in _____



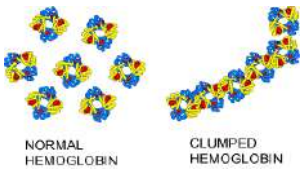


- Treatments for CF:
  - For respiratory symptoms:
    - _____
    - _____
    - _____
    - _____ in severe cases
  - For digestive symptoms:
    - Capsules containing _____

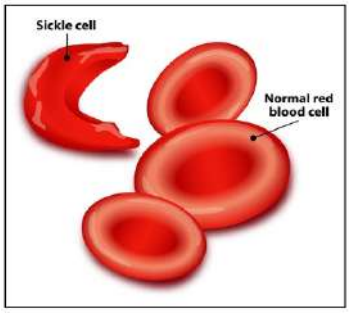
B) Sickle-Cell Anemia (_____)



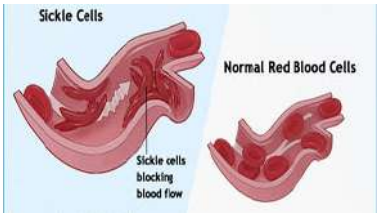
- The most common genetic disorder among _____
  - About 1 in 500 African Americans has sickle-cell anemia.
  - Carriers are said to have sickle-cell _____
- Caused by an abnormal gene on _____
  - The gene is for one of the polypeptide chains in _____, a protein found in _____ that is responsible for _____
  - Sickle-cell anemia causes hemoglobin to _____ within red blood cells, _____ from the normal biconcave disc to a sickle shape.



  - People with sickle-cell trait have some _____ but do not have the symptoms of sickle-cell disease



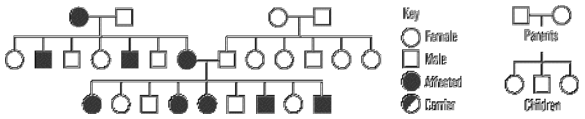
- Symptoms of Sickle-Cell Anemia:
  - Abnormal hemoglobin _____ as efficiently to cells as in healthy individuals
    - _____
    - _____
    - _____
  - Sickled red blood cells cannot move as easily through _____ as normal RBCs
    - Chronic _____, especially in _____
    - _____ to infections
    - _____



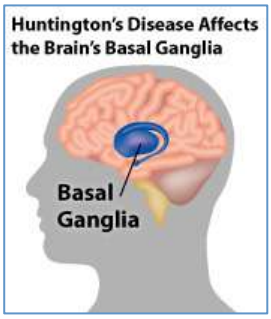
- Treatments for Sickle-Cell Anemia:
  - _____
  - _____
  - _____ that increase the oxygen-carrying capacity of red blood cells
  - Drugs that "switch on" the gene for _____ hemoglobin (normally switched off after birth)

- Heterozygote Superiority
  - Sickle-cell anemia is most common in areas of the world where _____ is prevalent
    - Malaria is caused by a parasite that _____
    - These parasites do not thrive in people with _____, so people with sickle-cell trait are _____ to malaria
  - People who are heterozygous for the cystic fibrosis allele may be more resistant to _____
  - When _____ have an advantage over people who are _____, it is called _____

C) Huntington's Disease



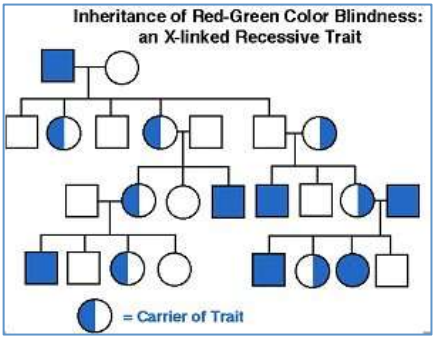
- Caused by an _____ (unlike most human genetic disorders)
  - Both men & women _____ to get the disorder
- Symptoms of Huntington's disease
  - Huntington's disease affects a person's _____
    - _____
    - _____
    - _____
    - _____
    - Loss of muscle coordination and ability to speak



  - Symptoms normally appear by _____
  - Huntington's disease is always _____
    - Death normally occurs within _____ after the onset of symptoms

3. Many genetic disorders are believed to be the result of _____:
   - _____ (Type I & II)
   - _____
   - _____
     - Bipolar disorder, schizophrenia
   - These are much more complicated to analyze than disorders caused by single genes

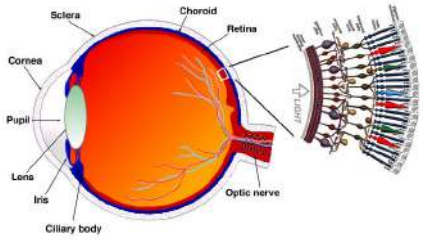
4. Sex-linked disorders are almost always caused by mutant alleles on the _____
   - _____
   - _____
     - Women can be _____, but men cannot
     - Ex: Homozygous normal female: X^BX^B
       Carrier female: X^BX^b
       Colorblind female: X^bX^b
       Normal male: X^BY
       Colorblind male: X^bY



A) Hemophilia is caused by an abnormal gene for _____

- Blood does not clot normally, so even a tiny cut can result in _____
- _____ is also a major concern
    - Most common around _____
- Hemophiliacs _____

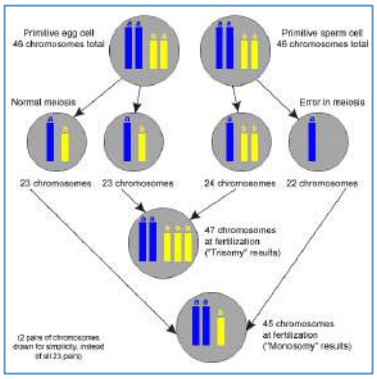
B) Red-green colorblindness is caused by an abnormal gene for _____



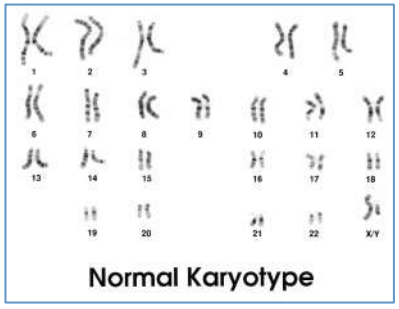
- The genes for both red and green photoreceptors are located on the X chromosome – colorblindness can result from recessive alleles for either one or both of these genes

5. Chromosome abnormalities are caused by mistakes made during meiosis

- May change the _____ or _____ of chromosomes in the gametes that are formed
    - _____ - the failure of a pair of chromosomes to separate during meiosis
        - Results in one gamete having too many chromosomes and another too few
        - _____ - a zygote gets 3 copies of a chromosome
        - _____ - a zygote gets only 1 copy of a chromosome



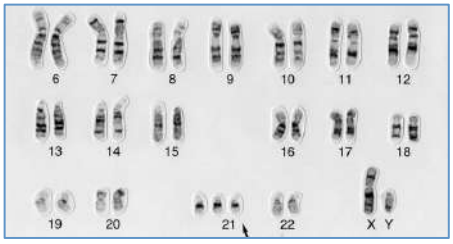
    - _____ is when a piece of one chromosome breaks off and attaches to a different chromosome
        - Often happens to 2 chromosomes at once

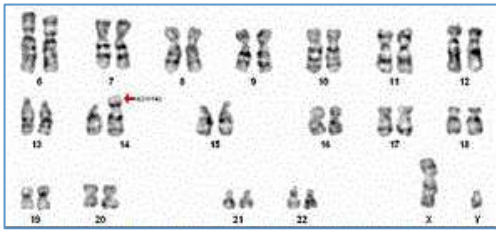


- Both nondisjunction and translocation can be detected in _____
    - Made from taking individual pictures of all of a human's chromosomes and matching up _____

A) **Down syndrome** – a genetic disorder that results from chromosome abnormality

- Nondisjunction – the person has an extra copy of _____
- Translocation – most of chromosome 21 breaks off during meiosis and fuses with another chromosome
    - Symptoms of Down syndrome:
        - Mild to severe _____
        - _____
        - _____
        - Susceptibility to _____ and _____





Section 2

1. _____ disabilities are different from genetic disorders
   - _____
   - Occur during _____

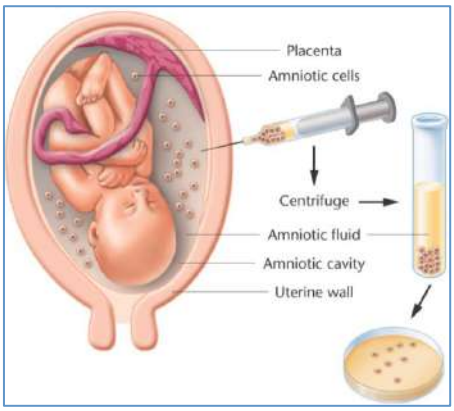
2. Both genetic and congenital disorders can often be detected _____

3. Genetic _____
   - Can help parents determine the _____ of their child being born with a genetic disorder
     - Genetic counselors study the _____ of both parents
       - Create _____ to trace the passage of traits
     - _____ analyze blood tests to determine if parents are _____ of certain genetic disorders
   - Usually can NOT determine whether or not a child will be born with a disorder, only the **probability**

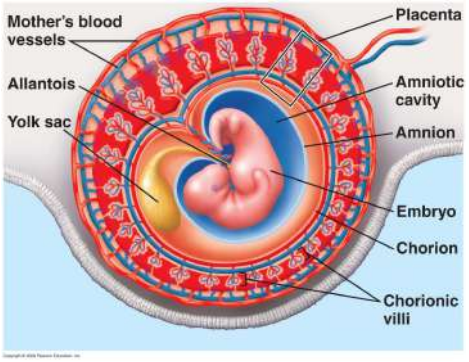
4. Two main ways to diagnose genetic disorders:
   - Analysis of _____
     - _____
     - _____
   - _____ techniques:
     - _____
     - _____

A) Amniocentesis

- _____ is the fluid that surrounds a fetus inside the uterus
  - Also contains fetal cells
- A sample of amniotic fluid is taken and cells are grown in a lab
  - Can be used to make a _____
    - Detects _____
  - Can be analyzed for _____
    - Detects _____
- Cannot be conducted until _____



B) Chorionic Villus Biopsy



- Chorionic villi are structures that help maximize the surface area for _____ between a mother and developing fetus (they are part of the _____)
- The villi develop from _____ and therefore contain the same _____ as the fetus & amniotic fluid
- A sample of these cells can be taken and analyzed as in amniocentesis
    - _____
    - _____
- Can be done as early as _____

C) Ultrasonography

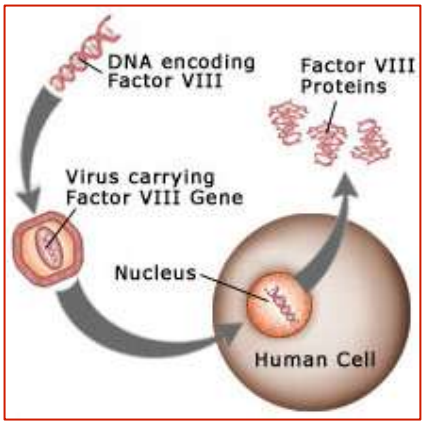
- Uses high-frequency _____ which bounce off of tissue
    - Depending on the _____ of the tissue, the waves "echo" back at different _____ and are used to produce a computerized image called an _____
- Used in most pregnancies to detect _____
- Used with amniocentesis to _____
- Can also help doctors detect abnormalities such as _____

D) Fetoscopy

- A _____ is made in a pregnant woman's _____
- An _____ is inserted through the incision
    - Has a _____ on the end that _____ on a monitor
    - _____ can be inserted through the endoscope tube to _____ _____

5. Developing cures for genetic disorders:

A) Gene therapy



- Introducing _____ into the cells of people with _____
    - Using _____ _____
    - Enclosing alleles in _____, which are taken into the cell by _____
- Currently these are still _____ and have had _____