Chapter 11 – Human Genetic Disorders – CP Biology

1. Major types of genetic disorders:
   • __________________________
     o __________________________
     o __________________________
   • __________________________
   • __________________________

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

A) Cystic Fibrosis
   • CF is the most common genetic disorder among __________________________
     o ~1 in 2500 white infants in the US are born with CF (4-5 born each day)
     o It is estimated that 1 in 20 white people is a carrier of the CF allele
   • Caused by an abnormal gene on __________________________
     o The gene is for a __________________________ that uses ________________
       to regulate the movement of sodium (Na⁺) and chloride (Cl⁻) ions into and out of cells
     o In healthy individuals, the normal protein __________________________
       • Keeps mucus thin and __________________________
     o With CF, not enough Cl⁻ ions are pumped out of cells
       • __________________________ in airways & pancreatic ducts
   • Symptoms of CF:
     o Buildup of mucus in __________________________
       • Difficulty __________________________
       • __________________________
     o Blocks __________________________ (produced by the pancreas) from entering the intestine
       • __________________________
     o Abnormal Na⁺ transport also results in __________________________
• Treatments for CF:
  o For respiratory symptoms:
    ▪ ____________________
    ▪ ____________________
    ▪ ____________________
    ▪ ____________________ in severe cases
  o For digestive symptoms:
    ▪ Capsules containing ____________________

B) Sickle-Cell Anemia (__________________________)
• The most common genetic disorder among ______________________
  o About 1 in 500 African Americans has sickle-cell anemia.
  o Carriers are said to have sickle-cell __________
• Caused by an abnormal gene on ______________________
  o The gene is for one of the polypeptide chains in ____________, a
    protein found in ________________ that is responsible for
    __________________________
  o Sickle-cell anemia causes hemoglobin to __________ within red blood
    cells, _____________________________ from the normal
    biconcave disc to a sickle shape.
  o People with sickle-cell trait have some __________________________
    but do not have the symptoms of sickle-cell disease
• Symptoms of Sickle-Cell Anemia:
  o Abnormal hemoglobin _____________________________ as
    efficiently to cells as in healthy individuals
    ▪ __________________
    ▪ __________________
    ▪ __________________
  o Sickled red blood cells cannot move as easily through __________________ as normal RBCs
    ▪ Chronic __________, especially in __________
    ▪ _____________________________ to infections
    ▪ __________________
• Treatments for Sickle-Cell Anemia:
  o ____________________
  o ____________________
  o _________ that increase the oxygen-carrying capacity of red blood cells
  o 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^B^B
    - Carrier female: X^B^x
    - Colorblind female: X^b^x
    - Normal male: X^B^Y
    - Colorblind male: X^b^Y
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
     o Genetic counselors study the _________________ of both parents
     ▪ Create _________________ to trace the passage of traits
     o _________________ 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 _________________
     o _________________
     o _________________
   • _________________ techniques:
     o _________________
     o _________________

A) Amniocentesis
   • _________________ is the fluid that surrounds a fetus inside the uterus
     o Also contains fetal cells
   • A sample of amniotic fluid is taken and cells are grown in a lab
     o Can be used to make a _________________
     ▪ Detects _________________
     o 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 ____________________________________________________________________________