Genetic Mutations

Indicator 4.8: Compare the consequences of mutations in body cells with those in gametes.
Agenda

• Warm UP: What is a mutation? Body cell? Gamete?

• Notes on Mutations
• Karyotype Web Activity
What is the difference in a body cell and gamete?

**Body Cell** – also known as somatic cell. Any cell other than sex cells. They are diploid.

Examples: skin, liver, blood cells

**Gametes** – also known as sex cells. They are haploid.

Examples: sperm and egg

Quick Review
Mutations
1) A mutation is the alteration of an organism’s DNA. It can range from a change in one base pair to the insertion or deletion of large segments of DNA.
2) Mutations are caused by a malfunction during meiosis or from exposure to a mutagen.

3) A mutagen is a physical or chemical agent that causes a mutation.
Mutations

4. Most mutations are automatically repaired and have no effect on the organism.

5. **Enzymes (proteins)** help with repairs.
1. If the mutation is not repaired the cell becomes a mutant cell and the mutation (altered chromosome or gene) is passed down to each daughter cell made during mitosis or meiosis.
2) If the mutant cell is a **body cell**:

a. **Daughter cells** will be affected by the altered DNA

b. The mutation will **not** be passed to the **offspring** of the organism.

c. Body cell mutations contribute to **the aging process and the development of many types of cancer**

Mutant Cells
2) If the mutant cell is a *gamete* (sex cell):

a. The altered DNA will be passed onto the *embryo* and may be passed to *future generations*.

b. Gamete cell mutations can result in *genetic disorders*.
1) A gene mutation is **a mutation that affects a single gene**

2) The basis of **sickle-cell disease** is the mutation of a single base pair in the gene that codes for **one** of the proteins of **hemoglobin**.

3) Other examples include **Tay-Sachs disease**, **Huntington’s disease**, **cystic fibrosis**, or **albinism**.

**Genetic Disorders: Gene Mutations**
Sickle Cell Anemia

- Recessive
- Defective Hemoglobin on RBCs
- Anemia (loss of blood cells)
- Damage to brain, heart, lungs
- Primarily in African Americans; 1/10 in US is a carrier

http://www.pbs.org/wgbh/evolution/library/01/2/quicktime/l_012_02.html
Tay-Sachs disease

- Recessive
- Lack of enzyme hexosaminidase A (hex A), which breaks down fatty acids in brain in nervous tissue
- Symptoms appear at 4-6 months
- Death by 5 years
- Found primarily in those descendants of Ashkenazi Jews
  - 1/30 American Jews carry the gene
From the US National Library of Medicine...

- Tay-Sachs disease is caused by a defective gene on chromosome 15. When both parents carry the defective Tay-Sachs gene, a child has a 25% chance of developing the disease. The child must receive two copies of the defective gene -- one from each parent -- in order to become sick. If only one parent passes the defective gene to the child, the child is called a carrier. He or she won't be sick, but will have the potential to pass the disease to his or her own children.
Huntington’s disease

- Autosomal- Dominant
- Lethal
- Begins around ages 35-45
- First symptoms; mild forgetfulness and irritability
- Lose control over muscles
- Genetic Marker: short section of DNA that has a close association with a known gene
  - Presence of gene marker can indicate the presence of Huntington’s allele
  - People with marker have 90% chance of developing Huntington’s
  - Gene Marker and gene so close on chromosome rarely separated by crossing over during meiosis
From the Mayo Clinic…

- Huntington's disease is an inherited disease that causes the progressive breakdown (degeneration) of nerve cells in the brain. Huntington's disease has a broad impact on a person's functional abilities and usually results in movement, thinking (cognitive) and psychiatric disorders.
- Most people with Huntington's disease develop signs and symptoms in their 40s or 50s, but the onset of disease may be earlier or later in life.
- Medications are available to help manage the symptoms of Huntington's disease, but treatments can't prevent the physical, mental and behavioral decline associated with the condition.
Cystic Fibrosis

- Recessive
- Point mutation stops production of a protein in the lungs and pancreas
  - Prevents cells from transporting Cl- ions out of the cell
- Lung Congestion
  - Abnormally thick mucus lining in lungs
  - Chronic Bacterial Infections (pneumonia)
  - Treated with antibiotics, lung transplant, and new genetic engineering treatments
Cystic fibrosis is a disease passed down through families that causes thick, sticky mucus to build up in the lungs, digestive tract, and other areas of the body. It is one of the most common chronic lung diseases in children and young adults. It is a life-threatening disorder.

Millions of Americans carry the defective CF gene, but do not have any symptoms. That's because a person with CF must inherit two defective CF genes -- one from each parent. An estimated 1 in 29 Caucasian Americans have the CF gene. The disease is the most common, deadly, inherited disorder affecting Caucasians in the United States. It's more common among those of Northern or Central European descent.
Albinism

• Albinism occurs when one of several genetic defects makes the body unable to produce or distribute melanin, a natural substance that gives color to your hair, skin, and iris of the eye.

• The defects may be passed down through families.
From the US National Library of Medicine...

- Albinism occurs when one of several genetic defects makes the body unable to produce or distribute melanin, a natural substance that gives color to your hair, skin, and iris of the eye.
- The defects may be passed down through families.
- There are two main types of albinism:
  - Type 1 albinism is caused by defects that affect production of the pigment, melanin.
  - Type 2 albinism is due to a defect in the "P" gene. People with this type have slight coloring at birth.
- The most severe form of albinism is called oculocutaneous albinism. People with this type of albinism have white or pink hair, skin, and iris color, as well as vision problems.
Genetic Disorders: Chromosomal Mutations

1) Definition: mutation affects a group of genes or an entire chromosome.

2) Nondisjunction: when chromosomes do not separate properly during anaphase and the result is an abnormal number of chromosomes.

Remember:
There should be 2 of every Chromosome!
Genetic Disorders: Chromosomal Mutations

- **Inversion**
  - Example: A B C D E F

- **Duplication**
  - Example: A B C D D E F

- **Deletion**
  - Example: A B D E F

- **Insertion**
  - Example: A B C D L E F

- **Translocation**
  - Example:
    - Initial: A B C D E F
    - Final: A B C o p q
Chromosomal Mutations

3) Examples of abnormalities in humans due to nondisjunction of sex chromosomes:

1) Klinefelter’s syndrome (males)
2) Turner’s syndrome (females)
Klinefelter’s Syndrome

• In males
  • Trisomy of sex chromosomes; XXy male
  • Feminine Characteristics, Infertile
  • George Washington?
    • No Children – Sterile?
    • Dental Problems
    • Height – Very tall for generation
    • Still Inconclusive
Turner’s Syndrome

- In females
- Monosomy of Sex Chromosomes; XO female
- Infertile
- Dwarfism
- Overweight
- Some mental retardation
- Webbed Neck
- Develop as females
Chromosomal Mutations

3) Examples of abnormalities in humans due to nondisjunction of autosomal chromosomes:

1) Downs syndrome
- Trisomy 21
- Mild to severe mental retardation
- Distinct Facial Features
- Heart Defects
- Fingerprints – Sworl
- Most Common Birth Defect – 1/700 births
- Mother’s Age over 40 – 1/80
  - Problems during Oogenesis
**Karyotype**: chromosomes are stained and photographed under the microscope, cut from photo and arranged by size and shape; can detect chromosomal abnormalities.
Beneficial Mutations

1) In some cases mutations are **beneficial** to organisms.

2) Beneficial mutations are **changes that may be useful to organisms in different or changing environments**.

3) The phenotypic results of beneficial mutations (what they look like): **those that are favored by natural selection & increase in a population**
Now...

Work on Web Activity with Karyotypes
Finish Potato Head Genetics

Quiz on Heredity Tomorrow

• Study Guide due on Thursday!
Agenda 4.09.14

Practice Test
  – Review Meiosis and Heredity

Quiz on Heredity

• Study Guide due on Thursday/Tomorrow!
Do now…

Work on your BABY DRAGON!

• Draw and Label genotype and phenotype (Aa; no chin spike)
• Complete your 10 questions
• DUE on Wednesday with TEST

• Study Guide due on Tuesday!
MUTATIONS - alteration of organism’s DNA (one base pair or large segment)
Can result from mutagens
CONCEPT MAP

In Body Cells- altered DNA affects daughter cells but will not be passed to offspring.
CONCEPT MAP

In Gametes - altered DNA transmitted to embryo and passed to offspring

Result in: Genetic disorders through gene mutations or chromosomal mutations
Nondisjunction - chromosomes failed to separate correctly during meiosis. Some cells have more chromosomes while others have less than normal amount.
Beneficial - changes that may be useful to organism in different or changing environments. Result in favored phenotypes by natural selection.
**Karyotype:** chromosomes are stained and photographed under the microscope, cut from photo and arranged by size and shape; can detect chromosomal abnormalities
Observations?
Autosomes vs. Sex?