Genetics: Part IV

Genetic Disorders
What has gone wrong?
c. Certain human genetic disorders can be attributed to the inheritance of single gene traits or specific chromosomal changes, such as nondisjunction.

*Illustrative examples:* • Sickle cell anemia
• Tay-Sachs disease
• Huntington’s disease
• X-linked color blindness
• Trisomy 21/Down syndrome
• Klinefelter’s syndrome
3.A.4.b Some traits are determined by genes on sex chromosomes.

*Illustrative examples*

- Sex-linked genes on sex chromosome (X in humans)
- In mammals and flies, the Y chromosome is very small and carries few genes
Small change, Big problem

(a) Normal amino acid sequence

Thr 4
Pro 5
Glu 6
Glu 7

(b) Single change in amino acid sequence

Thr 4
Pro 5
Val 6
Glu 7

Normal red blood cells

Sickled red blood cells

Figure 3-13 Biological Science, 2/e

© 2005 Pearson Prentice Hall, Inc.
Sex Linked Traits

• Traits carried on the sex chromosomes are said to be sex linked.
• In humans, most sex-linked traits are carried on the X chromosome.
• Sex-linked traits are expressed more often in human males than females.
Example of Sex Linked Trait: Hemophilia
What is the probability that the son of a carrier female and a normal male will have hemophilia?
What is the probability that the son of a carrier female and a normal male will have hemophilia?

<table>
<thead>
<tr>
<th></th>
<th>$X^H$</th>
<th>$Y$</th>
</tr>
</thead>
<tbody>
<tr>
<td>$X^H$</td>
<td>$X^HX^H$</td>
<td>$X^HY$</td>
</tr>
<tr>
<td>$X^h$</td>
<td>$X^hX$</td>
<td>$X^hY$</td>
</tr>
</tbody>
</table>
Imagine a genetic counselor working with a couple who have just had a child who is suffering from Tay-Sachs disease. Neither parent has Tay-Sachs, nor does anyone in their families. Which of the following statements should this counselor make to this couple?

a. “Because no one in either of your families has Tay-Sachs, you are not likely to have another baby with Tay-Sachs. You can safely have another child.”

b. “Because you have had one child with Tay-Sachs, you must each carry the allele. Any child you have has a 50% chance of having the disease.”

c. “Because you have had one child with Tay-Sachs, you must each carry the allele. Any child you have has a 25% chance of having the disease.”

d. “Because you have had one child with Tay-Sachs, you must both carry the allele. However, since the chance of having an affected child is 25%, you may safely have thee more children without worrying about having another child with Tay-Sachs.”
Example of Sex Linked Trait: Colorblindness

**Normal**

**Protonopia**
Color blindness

Draw a Punnett Square to show how a color blind male could produce a family containing colorblind females.
Is your Punnett Square like this one?
Nondisjunction

Meiosis I

Nondisjunction
Nondisjunction

First division

Nondisjunction at second division

\( n + 1 \)

\( n - 1 \)

\( n \)
Trisomy 21
Down syndrome patients have noticeable traits.
Genetic Pedigree

First Generation

Second Generation

U

V

W

Third Generation

X

Y

Z

Key

- ○ = normal female
- □ = normal male
- ● = affected female
- ■ = affected male
- ○ = carrier female
- ■ = carrier male
Huntington’s Disease: A Late-Onset Lethal Disease

• Huntington’s disease is a degenerative disease of the nervous system
• The disease has no obvious phenotypic effects until the individual is about 35 to 40 years of age
• Once the deterioration of the nervous system begins the condition is irreversible and fatal
Huntington’s affects people after reproduction age so it continues to be maintained in a population.
Envision a family in which the grandfather, age 47, has just been diagnosed with Huntington’s disease. His daughter, age 25, now has a 2-year-old baby boy. No one else in the family has the disease. What is the probability that the daughter will contract the disease?

A. 0%  
B. 25%  
C. 50%  
D. 75%  
E. 100%
NONNUCLEAR INHERITANCE
• 3.A.4.c. Some traits result from nonnuclear inheritance

• 1. Chloroplasts and mitochondria are randomly assorted to gametes and daughter cells; thus traits determined by chloroplasts and mitochondrial DNA do not follow simple Mendelian rules.

• 2. In animals, mitochondrial DNA is transmitted by the egg and not by the sperm; as such, mitochondrial-determined traits are maternally inherited.
A. Nuclear DNA is inherited from all ancestors.

B. Mitochondrial DNA is inherited from a single lineage.
Mitochondrial Inheritance

• Sometimes referred to as Maternal Inheritance

• More than 40 known human disorders are attributed to mitochondrial inheritance.

• Links have been made between mtDNA and diabetes, certain cancers and aging just to name a few.
• 3.A.3.d
• Many ethical, social and medical issues surround human genetic disorders.
• Illustrative examples
  – Reproduction issues
  – Civic issues such as ownership of genetic information, privacy, historical context
For people at risk of Huntington's disease, deciding whether or not to have a genetic test is a difficult, and very personal decision. There is no right or wrong decision.
Created by:

Debra Richards
Coordinator of K-12 Science Programs
Bryan ISD
Bryan, TX