Autosomal Recessive, Autosomal Dominant, and X-linked Recessive
Autosomal Recessive Disorders

**CYSTIC FIBROSIS BY THE NUMBERS**

1 in 31

Americans are silent carriers of cystic fibrosis

1,000

Babies born each year with cystic fibrosis

25%

Chance of having a baby with cystic fibrosis if both parents are carriers
Friedreich's Ataxia
Anyone remember this guy? Who he was looking for?
Autosomal Dominant Disorders
Sex-linked Genetic Disorders
Carrier Parent

A  a

AA  Aa  aa
Affected Child  Carrier Child  Normal Child

A Recessive disorder

A Dominant disorder

H  h

Hh  HH

H  H

H  H

H = “Healthy”

D  d

Dd  dd

D  Dd  dd

D = “Disease”

Notice there are no alleles on the Y

One male offspring will be affected

1/2 of sons unaffected

1/2 of sons have hemophilia

daughters all unaffected
**Rules of Inheritance**

**Autosomal Recessive**
- Appears in both sexes with equal frequency
- Trait tend to skip generations
- Affected offspring are usually born to unaffected parents
- When both parents are heterozygous, approx. 1/4 of the progeny will be affected
- Appears more frequently among the children of consanguine marriages

**X-Linked Recessive**
- More males than females are affected
- Affected sons are usually born to unaffected mothers, thus the trait skips generations
- Approximately 1/2 of carrier mothers' sons are affected
- It is never passed from father to son
- All daughters of affected fathers are carriers

**Rules of Inheritance**

**Autosomal Dominant**
- Appears in both sexes with equal frequency
- Both sexes transmit the trait to their offspring
- Does not skip generations
- Affected offspring must have an affected parent unless they possess a new mutation
- When one parent is affected (het.) and the other parent is unaffected, approx. 1/2 of the offspring will be affected
- Unaffected parents do not transmit the trait
### Clues for Autosomal Inheritance

<table>
<thead>
<tr>
<th>Recessive</th>
<th>Dominant</th>
</tr>
</thead>
<tbody>
<tr>
<td>• individual expressing trait has 2 normal parents</td>
<td>• every affected person has at least one affected parent</td>
</tr>
<tr>
<td>• two affected parents can not have an unaffected child</td>
<td>• each generation will have affected individuals</td>
</tr>
</tbody>
</table>

### Clues for Sex-linked Inheritance

<table>
<thead>
<tr>
<th>Recessive</th>
</tr>
</thead>
<tbody>
<tr>
<td>• no father-to-son transmission</td>
</tr>
<tr>
<td>• predominantly males affected</td>
</tr>
<tr>
<td>• trait my skip generations</td>
</tr>
</tbody>
</table>

1. For each of the pedigrees below, identify the mode of inheritance and provide at least 2 reasons for your choice.

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**Pedigree a**

- **Pedigree c**

- **Pedigree b**

- **Pedigree d**
2. Below is a pedigree for an inherited lung disease. Provide the genotypes of each of the individuals marked with lower case letters.
3. Below is a pedigree for an inherited brain disease. Provide the genotypes of each of the individuals marked with lower case letters.
Pedigree & Genotyping

**Pedigree A**

1. *Which type* of inheritance pattern is likely based on the pedigree of Neurofibromatosis (NF), a genetic skin condition? *Based on what reasoning?*
   - Autosomal Recessive
   - Autosomal Dominant
   - X-linked recessive

2. For the following genotypes, what would be the appropriate phenotypes?

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Phenotype:</th>
</tr>
</thead>
<tbody>
<tr>
<td>NN</td>
<td>Affected with NF, Healthy, Incompatible with life</td>
</tr>
<tr>
<td>Nn</td>
<td></td>
</tr>
<tr>
<td>nn</td>
<td></td>
</tr>
</tbody>
</table>

3. Write the genotypes for at least 7 individuals next to the symbol.

4. Calculate the risk for persons 7 & 8 to have another affected child.

5. Extension: How did individual #6 become affected with NF? Hint: This actually occurs in 50% of cases!
Pedigree B

1. Which type of inheritance pattern is likely based on the pedigree of CIP (Congenital Insensitivity to Pain), a genetic condition? Based on what reasoning?
   - Autosomal Recessive
   - Autosomal Dominant
   - X-linked recessive

2. For the following genotypes, what would be the appropriate phenotypes?

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Phenotype:</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Affected with CIP, Healthy (no CIP gene), Healthy CIP Carrier</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Phenotype:</th>
</tr>
</thead>
<tbody>
<tr>
<td>NN</td>
<td></td>
</tr>
<tr>
<td>Nn</td>
<td></td>
</tr>
<tr>
<td>nn</td>
<td></td>
</tr>
</tbody>
</table>

3. Write the genotypes for at least 7 individuals next to their symbols.

4. Calculate the risk for persons III-4 & III-5 to have another affected child.

5. Extension: What is the chance that IV-5 is a carrier of the disease? Assume IV-5 is healthy and shows no symptoms of the disorder. Hint: the punnett square above can be used for interpretation.
Pedigree C

1. Which type of inheritance pattern is likely based on the pedigree of Hemophilia, a genetic condition? Based on what reasoning?

- Autosomal Recessive
- Autosomal Dominant
- X-linked recessive

2. For the following genotypes, what would be the appropriate phenotypes?

<table>
<thead>
<tr>
<th>Genotype:</th>
<th>Phenotype:</th>
</tr>
</thead>
<tbody>
<tr>
<td>X'X'</td>
<td>Hemophiliac, Healthy (non-carrier), Healthy Hemophilia Carrier</td>
</tr>
<tr>
<td>X'X</td>
<td></td>
</tr>
<tr>
<td>X'Y</td>
<td></td>
</tr>
<tr>
<td>X'Y</td>
<td></td>
</tr>
</tbody>
</table>

3. Write the genotypes for the following persons: Victoria, Albert, Alice of Hesse, Duke Leopold, Beatrice and Alice of Athlone.

4. Calculate the risk for persons Victoria and Albert to have another affected child (gender not known).
Genetics - X Linked Genes

**In fruit flies, eye color is a sex linked trait. Red is dominant to white.**

1. What are the sexes and eye colors of flies with the following genotypes?

   \[ X^R X^r \quad X^R Y \quad X^r X^r \quad X^R X^R \quad X^r Y \]

2. What are the genotypes of these flies:

   white eyed, male \__________\ red eyed female (heterozygous) \__________\n
   white eyed, female \__________\ red eyed, male \__________\n
3. Show the cross of a white eyed female \( X^r X^r \) with a red-eyed male \( X^R Y \).
4. Show a cross between a pure red eyed female and a white eyed male. What are the genotypes of the parents:

___________ and ___________

How many are:

white eyed, male ____
white eyed, female ____
red eyed, male ____
red eyed, female ____

5. Show the cross of a red eyed female (heterozygous) and a red eyed male.

What are the genotypes of the parents?

_________ & _____________

How many are:

white eyed, male ____
white eyed, female ____
red eyed, male ____
red eyed, female ____

Math: What if in the above cross, 100 males were produced and 200 females. How many total red-eyed flies would there be? _______