Section 15.1
Human Chromosomes
I Can...

• **LS 3.3** I can compare different modes of inheritance (sex-linkage, codominance, incomplete dominance, multiple alleles, and polygenic traits).

• **LS 3.3** I can apply pedigree data to interpret various modes of genetic inheritance.

• **LS 3.3** I can use pedigree analysis of trait inheritance and to predict family member genotypes.
Key Questions

1. How are human karyotypes used?
2. What patterns of inheritance do human traits follow?
3. How can pedigrees be used to analyze human inheritance?
Vocabulary

• Genome
• Karyotype
• Sex chromosome
• Autosome
• Sex-linked gene
• Pedigree
Karyotypes

- **Genome** – the full set of genetic information that an organism carries in its DNA
- To see human chromosomes clearly, cell biologists photograph cells in mitosis
- A **karyotype** shows the complete diploid set of chromosomes grouped together in pairs, arranged in order of decreasing size
- Typical human cells
  - 46 chromosomes, 23 pairs
Sex Chromosomes

- XX = female (biological sex)
- XY = male (biological sex)
- 50/50 chance that a zygote will be male or female
  - All human egg cells are (23,X)
  - Human sperm cells can be (23,X) or (23,Y)

- X chromosome – has more than 1400 genes
- Y chromosome – has only about 158 genes
Autosomal Chromosomes

• The remaining 44 human chromosomes

• The complete human genome contains 46 chromosomes
  • 44 autosomes
  • 2 sex chromosomes

• Females = 46, XX
• Males = 46, XY
Transmission of Human Traits

- Simple dominance
- Codominance
- Multiple Alleles
- Sex-linked inheritance
- X-chromosome inactivation
Sex-Linked Inheritance

• A **sex-linked** gene is a gene located on a sex chromosome (X or Y)

• Example- colorblindness
  • Humans have 3 alleles responsible for colorblindness, all on the X chromosome
  • In males, a defective version of any one of these genes produces colorblindness
## Colorblindness

<table>
<thead>
<tr>
<th>Father (normal vision)</th>
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<tbody>
<tr>
<td>$X^C_Y$</td>
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<th>$X^C$</th>
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<tr>
<td>$X^C$</td>
<td>$X^C_Y$</td>
<td>$X^C_Y$</td>
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<tr>
<td>Daughter (normal vision)</td>
<td>$X^C_X^C$</td>
<td>$X^C_X^C$</td>
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<td>Son (normal vision)</td>
<td>$X^C_Y$</td>
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<tr>
<td>$X^C$</td>
<td>$X^C_Y$</td>
</tr>
<tr>
<td>Mother (carrier)</td>
<td>$X^C$</td>
</tr>
<tr>
<td>Daughter (carrier)</td>
<td>$X^C_X^C$</td>
</tr>
<tr>
<td>Son (colorblind)</td>
<td>$X^C_Y$</td>
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</tbody>
</table>
X-Chromosome Inactivation

- If just one X chromosome is enough for cells in males, how does the cell “adjust” to the extra X chromosome in female cells?
- One X chromosome can be switched “off”
- Forms a condensed region in the nucleus called a Barr body

- Example- calico cats
  - Females are white with orange and black spots
  - Males are white with spots of one color
Human Pedigrees

• Chart that shows the relationships within a family
• Shows the presence or absence of one human trait
• Used to determine whether a trait is...
  • Dominant or recessive
  • Autosomal or sex-linked
A circle represents a female.
A square represents a male.
A horizontal line connecting a male and a female represents a marriage.
A shaded circle or square indicates that a person expresses the trait.
A circle or square that is not shaded indicates that a person does not express the trait.

存在的白色前额毛 = 蓝色（蓝色）
未存在的前额毛 = 白色
未存在的前额毛 = 白色

White forelock

1 = presence of white forelock trait
0 = no forelock trait
Pedigree Analysis

**Autosomal or sex-linked?**

- **Autosomal**
  - Affects males and females the same

- **Sex-linked on the X**
  - More common in males

- **Sex-linked on the Y**
  - In males ONLY

**Dominant or recessive?**

- **Dominant**
  - Trait is seen in all generations
  - No carriers

- **Recessive**
  - Trait “skips” generations
  - Could have carriers
Section 15.1 Exit Ticket

1. What is a karyotype? What information does it depict?
2. What are the conventions of a pedigree?
3. How can pedigree analysis be used to determine if a genetic trait is X-linked dominant? X-linked recessive? Autosomal dominant? Autosomal recessive?
The End ☺️