8. On chromosome 3 of corn there is a dominant gene (G) which together with the dominant gene (R) on chromosome 9 produces colored aleurone (i.e. G-R- is colored). All other genetic combinations produce colorless aleurone. Two homozygous colorless strains are crossed to produce an all colored F₁.

a. What are the genotypes of the two homozygous colorless parental strains? (4)

b. What is the genotype of the colored F₁? (2)

c. What phenotypic proportions are expected in the F₂? (4)

9:7 (colored:colorless)

“Proof” of the Sutton-Boveri Chromosome Theory of Inheritance

- Sex determination
- Meiotic nondisjunction
- Sex linkage
- Morgan 1910
- Bridges 1916
(a) X–Y system in mammals

(b) The X–0 system in certain insects

(c) The Z–W system in birds

Carothers (1913)
**Ascaris incurva**

**Female**
- 13 pair A + 8 pair X = 42

**Male**
- 13 pair A + 8X + 1Y = 35

Y bearing sperm  
13A + 1Y

X bearing sperm  
13A + 8X

Egg  
13A + 8X

---

**numeric balance**

<table>
<thead>
<tr>
<th>Chromosome composition</th>
<th>Genotypic composition</th>
<th>Ratio of X chromosomes to autosome combination</th>
<th>Sexual morphology</th>
</tr>
</thead>
<tbody>
<tr>
<td>20.2µm</td>
<td>1.0</td>
<td>Male</td>
<td></td>
</tr>
<tr>
<td>18.2µm</td>
<td>1.0</td>
<td>Female</td>
<td></td>
</tr>
<tr>
<td>18.1µm</td>
<td>1.0</td>
<td>Female</td>
<td></td>
</tr>
<tr>
<td>18µm</td>
<td>1.0</td>
<td>Female</td>
<td></td>
</tr>
<tr>
<td>17µm</td>
<td>1.0</td>
<td>Female</td>
<td></td>
</tr>
<tr>
<td>16µm</td>
<td>1.0</td>
<td>Female</td>
<td></td>
</tr>
<tr>
<td>15µm</td>
<td>1.0</td>
<td>Female</td>
<td></td>
</tr>
<tr>
<td>14µm</td>
<td>1.0</td>
<td>Female</td>
<td></td>
</tr>
<tr>
<td>13±µm</td>
<td>1.0</td>
<td>Female</td>
<td></td>
</tr>
</tbody>
</table>

**numerator : denominator genes**
Ascaris incurva

Female

• 13 pair A + 8 pair X = 42

Male

• 13 pair A + 8X + 1Y = 35

Y bearing sperm - 13A + 1Y

X bearing sperm - 13A + 8X

Egg - 13A + 8X

“Proof” of the Sutton-Boveri Chromosome Theory of Inheritance

- Sex determination
- Meiotic nondisjunction
- Sex linkage
- Morgan 1910
- Bridges 1916

Independent origin of chicken Z and human X chromosomes

**Figure 8.22**

Monosomy: 2N – 1

Trisomy: 2N + 1

<table>
<thead>
<tr>
<th>Aneuploid Conditions in Humans</th>
<th>Condition</th>
<th>Frequency</th>
<th>Syndrome</th>
<th>Characteristics</th>
</tr>
</thead>
<tbody>
<tr>
<td>Autosomal</td>
<td>Trisomy 18</td>
<td>1/8,000</td>
<td>Edward</td>
<td>Mental and physical retardation, facial abnormalities, extreme muscle tone, early death</td>
</tr>
<tr>
<td></td>
<td>Trisomy 13</td>
<td>1/15,000</td>
<td>Patau</td>
<td>Mental and physical retardation, wide variety of defects in organs, large triangular nose, early death</td>
</tr>
<tr>
<td>Sex Chromosomal</td>
<td>XXX (male)</td>
<td>1/1,500</td>
<td>Klinefelter</td>
<td>Sexual immaturity (big sperm), breast swelling</td>
</tr>
<tr>
<td></td>
<td>XXY (male)</td>
<td>1/1,000</td>
<td>Jacobs</td>
<td>Tall</td>
</tr>
<tr>
<td></td>
<td>XXX (female)</td>
<td>1/1,500</td>
<td>Triple X</td>
<td>Tall and thin, menstrual irregularity</td>
</tr>
<tr>
<td></td>
<td>XO (female)</td>
<td>1/15,000</td>
<td>Turner</td>
<td>Short stature, webbed neck, sexual underdevelopment</td>
</tr>
</tbody>
</table>
“Proof” of the Sutton-Boveri Chromosome Theory of Inheritance

- Sex determination
- Meiotic nondisjunction
- Sex linkage
- Morgan 1910
- Bridges 1916

Sex Linkage

- X linkage
- Y linkage (holandric genes)
- X and Y linkage

Sex Linkage

- X linkage
- Y linkage (holandric genes)
- X and Y linkage

Duchenne muscular dystrophy
Males are said to be hemizygous for X-linked traits; the term pseudodominance is used when a recessive allele is expressed in the phenotype even though only a single copy of the allele is present (as occurs in the cross on the right). The term is today primarily used when the deletion of the dominant allele in a heterozygote produces a recessive phenotype individual.

Sex Linkage

- X linkage
- Y linkage (holandric genes)
- X and Y linkage

Hairy ears – a holandric gene?
Bobbed bristles in *Drosophila*

P1 x P2:

\[ X^{bb}X^{bb} \times X^{bb+}Y^{bb+} \rightarrow X^{bb}X^{bb+} + X^{bb+}Y^{bb+} \]

(Comment: F1 are all wild-type)

F1 x F1:

\[ X^{bb}X^{bb+} \times X^{bb+}Y^{bb+} \rightarrow \]

F2:

\[ \frac{1}{4} X^{bb}X^{bb} + \frac{1}{4} X^{bb}X^{bb+} + \frac{1}{4} X^{bb+}Y^{bb+} + \frac{1}{4} X^{bb+}Y^{bb+} \]

(Comment: Because the F2 results are a “3:1” ratio, x-and-y linked traits are said to have a “pseudoautosomal pattern” of inheritance)

---

Y chromosome:

PAR, pseudoautosomal region

NRY, nonrecombining region

---

10. In the pedigree shown below for a trait determined by a single gene (affected individuals are shown in black), state whether it would be possible or impossible to be inherited in each of the following ways. If impossible, cite a specific mating to help you in explaining your conclusion.

A. autosomal recessive
B. X-linked recessive
C. autosomal dominant, complete penetrance and expressivity
D. X-linked dominant, complete penetrance and expressivity

---

Shared in common 320 MYA

Common ancestor of humans/rhesus monkey 25 MYA

Ancestral X genes today
10. In the pedigree shown below for a trait determined by a single gene (affected individuals are shown in black), state whether it would be possible or impossible to be inherited in each of the following ways. If impossible, cite a specific mating to help you in explaining your conclusion.

A. autosomal recessive – Only this is possible
B. X-linked recessive
C. autosomal dominant, complete penetrance and expressivity
D. X-linked dominant, complete penetrance and expressivity

6. If two parents who are both type A and have normal vision produce a son who is color-blind and type O, what is the probability that their next child will be a female who has normal vision and is blood type A?

Half of their children will be daughters with normal vision. You would expect ¼ to be type A.

½ x ¾ = 3/8
“Proof” of the Sutton-Boveri Chromosome Theory of Inheritance

- Sex determination
- Meiotic nondisjunction
- Sex linkage
- Morgan 1910
- Bridges 1916

Doncaster and Raynor (1906)

*Abraxas* – a moth

male x female → progeny

dark       light       all dark

light       dark       light ♀, dark ♂

Morgan (1910)

Cross A

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Parentage</th>
<th>Sex</th>
</tr>
</thead>
<tbody>
<tr>
<td>1/2 red ♀</td>
<td>red ♂</td>
<td>red</td>
</tr>
<tr>
<td>1/2 red ♀</td>
<td>white ♂</td>
<td>white</td>
</tr>
</tbody>
</table>

F₁

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Parentage</th>
<th>Sex</th>
</tr>
</thead>
<tbody>
<tr>
<td>1/2 red ♀</td>
<td>red ♂</td>
<td>red</td>
</tr>
<tr>
<td>1/2 red ♀</td>
<td>white ♂</td>
<td>white</td>
</tr>
</tbody>
</table>

F₂

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Parentage</th>
<th>Sex</th>
</tr>
</thead>
<tbody>
<tr>
<td>1/4 red ♀</td>
<td>(2459)</td>
<td>red</td>
</tr>
<tr>
<td>1/4 red ♀</td>
<td>(1011)</td>
<td>red</td>
</tr>
<tr>
<td>1/4 white ♀</td>
<td>(782)</td>
<td>white</td>
</tr>
<tr>
<td>1/4 white ♀</td>
<td>(86)</td>
<td>white</td>
</tr>
</tbody>
</table>

Cross B

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Parentage</th>
<th>Sex</th>
</tr>
</thead>
<tbody>
<tr>
<td>1/2 red ♂</td>
<td>red ♂</td>
<td>red</td>
</tr>
<tr>
<td>1/2 white ♂</td>
<td>white ♂</td>
<td>white</td>
</tr>
</tbody>
</table>

F₁

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Parentage</th>
<th>Sex</th>
</tr>
</thead>
<tbody>
<tr>
<td>1/2 red ♀</td>
<td>red ♂</td>
<td>red</td>
</tr>
<tr>
<td>1/2 red ♀</td>
<td>white ♂</td>
<td>white</td>
</tr>
</tbody>
</table>

F₂

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Parentage</th>
<th>Sex</th>
</tr>
</thead>
<tbody>
<tr>
<td>1/4 red ♀</td>
<td>(129)</td>
<td>red</td>
</tr>
<tr>
<td>1/4 red ♀</td>
<td>(88)</td>
<td>red</td>
</tr>
<tr>
<td>1/4 white ♀</td>
<td>(132)</td>
<td>white</td>
</tr>
<tr>
<td>1/4 white ♀</td>
<td>(86)</td>
<td>white</td>
</tr>
</tbody>
</table>
Doncaster and Raynor (1906)

*Abraxas* – a moth

\[
\text{male} \times \text{female} \rightarrow \text{progeny}
\]

- dark \times light \rightarrow \text{all dark}
- light \times dark \rightarrow \text{light } \varphi, \text{dark } \sigma'

Doncaster and Raynor (1906)

\[
\text{male} \times \text{female} \rightarrow \text{progeny}
\]

- dark \times light \rightarrow \text{all dark}
- light \times dark \rightarrow \text{light } \varphi, \text{dark } \sigma'

Morgan (1910)

\[
\text{Cross A} \quad \begin{array}{c}
\text{P}_1 \quad \text{red} \quad \times \quad \text{white} \\
1/2 \text{ red } \\
1/2 \text{ red (2459)} \\
1/4 \text{ red (1011)} \\
1/4 \text{ white (782)} \\
\end{array} \\
\text{F}_1 \quad \begin{array}{c}
1/2 \text{ red} \\
1/2 \text{ white} \\
1/4 \text{ red (129)} \\
1/4 \text{ white (88)} \\
\end{array} \\
\text{F}_2 \quad \begin{array}{c}
1/4 \text{ red (132)} \\
1/4 \text{ white (86)} \\
\end{array}
\]

\[
\text{Cross B} \quad \begin{array}{c}
\text{P}_0 \quad \text{white} \quad \times \quad \text{red} \\
1/2 \text{ white} \\
1/2 \text{ white (86)} \\
\end{array} \\
\text{F}_1 \quad \begin{array}{c}
1/2 \text{ white} \\
1/2 \text{ red (129)} \\
1/4 \text{ red (88)} \\
1/4 \text{ white (86)} \\
\end{array} \\
\text{F}_2 \quad \begin{array}{c}
1/4 \text{ white (86)} \\
1/4 \text{ red (86)} \\
\end{array}
\]
Bridges (1916)

X-X' Pairing  

X-X (white) ♀ × X-Y (red) ♂

X-X' (red) ♀ × X-Y' (white) ♂

Regular offspring

Red sterile ♂ White ♀ × X-Y (red) ♂

Primary exceptions

Red ♂ White ♀ Red sterile ♂ White ♀

96% Regular  4% Secondary exceptions

X-Y Pairing

XX  XY

Y  X

X ←→ X  X ←→ Y

↓  ↓

XY  X  XX  Y

X  XY  X  XY
5. Hemophilia is a sex-linked recessive trait. A son born to phenotypically normal parents has Klinefelter Syndrome (XXY) and hemophilia, an X-linked recessive trait. In which parent and in which meiotic division did nondisjunction of the X chromosome most likely occur? Explain your reasoning.

Dad does not have the hemophilia allele so Mom must. A nondisjunction in the first meiotic division would have produced only X\textsuperscript{H}X\textsubscript{h} gametes whereas if it had occurred in the second division she could produce X\textsubscript{h}X\textsubscript{h} gametes which if combined with a Y-bearing sperm produce an X\textsubscript{h}X\textsubscript{h}Y male hemophiliac with Klinefelter Syndrome.

Epigenetic Inheritance

- Gene expression is modified but modification is not permanent over many generations

• Genetic imprinting
Epigenetic Inheritance

- Gene expression is modified but modification is not permanent over many generations

- Genetic imprinting
- Dosage compensation

Mary Lyon (1961) and Liane Russell (1961) hypothesis:
“heterochromatization”

Calico cat – What sex is this cat?

Are male Calico cats a possibility?
If all but one X chromosome is inactivated in humans then why aren’t XO and XXY individuals normal?

\[
\text{XO} = \text{XX} \\
\text{XXY} = \text{XY}
\]
If all but one X chromosome is inactivated in humans then why aren’t XO and XXY individuals normal?

XO  =  X
XY = XY

If all but one X chromosome is inactivated in humans then why aren’t XO and XXY individuals normal?

XO  =  XX
XXY = XY

**TABLE 7.1**

<table>
<thead>
<tr>
<th>Species</th>
<th>Sex Chromosomes in:</th>
<th>Females</th>
<th>Males</th>
<th>Mechanism of Compensation</th>
</tr>
</thead>
<tbody>
<tr>
<td>Fetal mammal</td>
<td>XX</td>
<td>XY</td>
<td></td>
<td>One of the X chromosomes in the somatic cells of females is inactivated.</td>
</tr>
<tr>
<td>Fetal mammal</td>
<td>XX</td>
<td>XY</td>
<td></td>
<td>The paternally derived X chromosome is inactivated in the somatic cells of females.</td>
</tr>
<tr>
<td>Marsupial mammal</td>
<td>XX</td>
<td>XY</td>
<td></td>
<td>The level of expression of genes on the X chromosome in males is increased 2-fold.</td>
</tr>
<tr>
<td>Drosophila melanogaster</td>
<td>XX</td>
<td>XY</td>
<td></td>
<td>The level of expression of genes on both X chromosomes in hermaphrodites is decreased to 50% levels compared to males.</td>
</tr>
</tbody>
</table>

*In C. elegans, an XX individual is a hermaphrodite, not a female.*