Ch. 20
Genes and Inheritance

How traits are passed from generation to generation

What are Genes?
- Each chromosome contains one very long DNA molecule
- Typically bears thousands of genes
- Genes carry our traits
- Genes are sections of DNA

What is heredity?
- Heredity – passing traits from parent to offspring
- The genes for certain traits are passed down in families from parents to children.
- For example,
  - parents with curly hair will have kids with curly hair
  - parents with short fingers will have kids with short fingers

Terms used in modern genetics
- Genes
  - Carry the instructions for making proteins
  - Each gene is found at a specific site on a chromosome
- Alleles
  - are different versions of a gene
  - Diploid cells (2n) have pairs of genes on homologous chromosomes

Genes carry our traits
- Genes
  - found at specific locations on a chromosome
- Alleles
  - are different versions of a gene

Genotype and phenotype
- Genotype – an individual’s genes
  - Allele “F” codes for freckles
- Phenotype – what an individual looks like
Genotype and phenotype

- What is her genotype?
- The gene for freckles has 2 alleles
  - The dominant allele = F
  - The recessive allele = f
- Chromosomes are found in pairs
- Possible genotypes:
  
<p>| | |</p>
<table>
<thead>
<tr>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>F</td>
<td>F</td>
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<tr>
<td>F</td>
<td>f</td>
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</tbody>
</table>

Examples of traits controlled by a single gene in humans

- These traits are determined by simple dominant-recessive inheritance
- Possible genotypes for someone with freckles?
- Without freckles?

Inheriting a trait

- If a man with short fingers marries a woman with long fingers, what genotypes and phenotypes will their children have?

<table>
<thead>
<tr>
<th>Dad</th>
<th>Mom</th>
</tr>
</thead>
<tbody>
<tr>
<td>F</td>
<td>f</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Dominant trait</th>
<th>Recessive trait</th>
</tr>
</thead>
<tbody>
<tr>
<td>FF or Ff</td>
<td>ff</td>
</tr>
</tbody>
</table>

Forming the gametes

- Gametes carry only one allele for each trait.

<table>
<thead>
<tr>
<th>Dad</th>
<th>Mom</th>
</tr>
</thead>
<tbody>
<tr>
<td>F</td>
<td>f</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Genotype = FF</th>
</tr>
</thead>
</table>

Gametes are haploid — only carry one of each chromosome

Fertilization

- When sperm and egg unite at fertilization, each contributes its allele.

<table>
<thead>
<tr>
<th>Fertilized egg</th>
</tr>
</thead>
<tbody>
<tr>
<td>Genotype = Ff</td>
</tr>
</tbody>
</table>

Phenotype?
Forming the gametes

- What if dad is \( FF \)?

\[
\text{Dad} \quad F \quad f
\]

\[
\text{Genotype} = FF
\]

\[
\text{meiosis} \quad F \quad f \quad F \quad f
\]

Then after fertilization ...

\[
\text{Dad} \quad \text{Genotype} = Ff \quad \text{F or f} \quad \text{F or f}
\]

\[
\text{Mom} \quad \text{Genotype} = \text{f or f} \quad \text{All f}
\]

\[
\text{Gametes:} \quad F \quad f \quad \text{Fertlized egg: genotype? phenotype?} \quad F or ff
\]

1 gene → 1 trait

- If neither parent has freckles, what will be the genotype of their offspring?

\[
\text{Offspring} = \text{ff}
\]

Inheritance (more complicated)

- If both parents are heterozygous for freckles, what will be the genotype of their offspring?

\[
\text{Freckles} \quad F \quad f \quad \text{Freckles} \quad F \quad f
\]

\[
\text{Gametes?} \quad F \quad f \quad \text{Gametes?} \quad F \quad f
\]

Punnett square analysis

- Grid used to predict the genotypes of the offspring.
- Shows all possible combinations of egg and sperm!

\[
\begin{array}{|c|c|c|}
\hline
\text{Genotype? Gametes?} & 1 & 2 \\
\hline
\text{Mom} \quad Ff & F & f & F & f \\
\hline
\text{Dad} \quad Ff & F & f & F & f \\
\hline
\end{array}
\]

\[
\text{Punnett square analysis}
\]

What are the genotypes of the offspring of two heterozygous freckled parents (\( Ff \))?

\[
\begin{array}{|c|c|}
\hline
\text{Gametes?} & 1 & 2 \\
\hline
\text{Ff} & F & f \\
\hline
\text{ff} & f & f \\
\hline
\end{array}
\]

\[
\text{Ratio: 1FF, 2 Ff, 1 ff}
\]

\[
\text{What are the phenotypes?}
\]
Now it’s your turn

- Tim and Jan both have free earlobes but their son does not. Show with a Punnett square how this can happen.

Recessive disorders

- Most human genetic disorders are recessive.
- Albinism
  - lack of melanin pigment in the eyes, skin and hair
  - affects mammals (including humans), fish, birds, reptiles and amphibians

Imagine you are a genetic counselor

- Cystic fibrosis
  - Most common lethal genetic disease in US
  - Symptoms: excessive secretion of a very thick mucus which interferes with breathing
  - Symptoms usually appear shortly after birth.

- A man and a woman who have a family history of CF are thinking of having a child.
- Genetic analysis showed they are both carriers of the defective gene
- What would be your advice?
- What is the probability that their child will have CF?

Dominant Disorders

- Achondroplasia is a common form of dwarfism.
- Caused by a mutation in the FGFR3 gene on chromosome 4
  - abnormal bone and cartilage formation
  - Just need 1 copy of mutated gene

Huntington disease: a dominant genetic disorder

- Caused by mutations in the HTT gene.
- Every individual who carries the allele gets the disorder
- Fatal: causes progressive deterioration of the brain
- Late age of onset: most people do not know they are affected until they are more than 30 years old

Recessive disorders

- Albino alligator
  - Genotype = mm

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Beyond simple inheritance

Most traits involve multiple genes

Skin color is determined by several genes

Incomplete Dominance

- The dominant allele codes for protein “H”
- A single dose of the protein gives an intermediate result

Incomplete Dominance

- Curly hair: hh
- Wavy hair: Hh
- Straight hair: HH

Multiple Alleles:

ABO blood groups

- Three alleles for the same gene
  - \( I^A \): Carbohydrate A on RBC
  - \( I^B \): Carbohydrate B on RBC
  - \( i \): neither A or B
- The \( I^A \) and \( I^B \) alleles exhibit codominance.
- Both alleles are expressed in the heterozygote.

Blood types and paternity tests

- A paternity suit is filed by a woman with Type O blood.
- The man accused of being the father has Type A blood.
- The child is type O.
- Could the man be the father? If he is, what must be his genotype?

Pleiotropy

- When a single gene has more than 1 effect

Marfan syndrome— an example of pleiotropy

- A mutation in the \( FBN1 \) gene
- Connective tissue defects
- Disproportionately long hands, a weak aorta, caved in breastbone, and other symptoms
Most traits are controlled by multiple genes

- At least 180 genes control how tall a person will grow

Some disorders are controlled by genes on the X chromosome

- Sex-linked inheritance involves genes located on a sex chromosome
  - Most are on the X chromosome.
  - X chromosomes contain nearly 2000 genes
  - Y chromosomes contain only 80 genes
  - Whether you’re male or female affects the pattern of inheritance
    - Why?

Red-green colorblindness

- X-linked disorder
  - due to a recessive allele on the X chromosome
- Red-green color blindness
  - The light-sensitive cells in the eyes don’t function properly.
  - Mostly found in males
  - Prevalence: 5-10% of males

A test for red-green colorblindness

Inheriting colorblindness

- A man with normal vision and a woman carrier have children. What is the chance that the couple will have a color-blind daughter? A color-blind son?
  - Man’s genotype
  - Man’s gametes
  - Woman’s genotype
  - Woman’s gametes

Sex-Linked Disorders

- Male pattern baldness
  - Largely sex-linked, but other genes are also involved
  - Gene is on the X chromosome
  - Passed from mother to son
  - Because the allele is recessive, a female needs two X chromosomes with the defect to show typical male pattern baldness

Inheriting baldness

- What is the chance that their sons will be bald?
  - What genotypes are possible for their daughters? Phenotypes?