Chemical Biology 03
Dec 4, 2009

Inheritance II
Unique steps for meiosis:
what do they accomplish?
Before we knew any of this...
Gregor Mendel dreamed up the exact same behavior for the mysterious (still abstract) units of heredity

1. Discrete genetic factors ("genes") exist and are passed unchanged from one generation to another

3. Each gene can exist in multiple forms ("alleles")

   \[
   \begin{align*}
   a & \quad A \\
   I^a & \quad I^b & \quad I^c
   \end{align*}
   \]

2. Each individual carries a pair of alleles for each gene
   - two alleles can be the same ("homozygous") \(aa\) \(AA\)
   - or different ("heterozygous") where one may be dominant and one recessive \(aA\)

5. During gametogenesis: alleles are segregated from each other: gametes carry only one allele \(A \leftrightarrow a\)

6. Alleles unite at random during fertilization

7. The alleles of different genes behave independently (independent assortment)
Mendel’s First Law of Segregation:

• Each individual carries a pair of alleles for each gene

• The two alleles segregate from one another during gametogenesis

• Alleles unite at random (one from each parent) during fertilization
Consider a cross:  
\[ Aa \times Aa \]

Fertilization is random!

\[
\begin{array}{c|c|c|c}
A & AA & Aa & \text{gametes} \\
\hline
a & Aa & aa & \text{offspring} \\
\end{array}
\]

\[
\frac{1}{4} \text{ homozygous} \\
\frac{1}{2} \text{ heterozygous} \\
\frac{1}{4} \text{ homozygous}
\]

Mendelian Ratio: 1/4 : 2/4 : 1/4
Predicting the outcome of a cross:

What is the probability of getting \( aa \)?

**Product Rule:**
Probability that two independent events will occur simultaneously = *product* of the probability of each event occurring on its own.

Use **Product Rule:** \( \frac{1}{2} \times \frac{1}{2} = \frac{1}{4} \) (and)
Predicting the outcome of a cross:

\[ Aa \times Aa \]

\[ \begin{array}{c|c|c|c}
  & A & a \\ \hline
A & AA & aA \\ \hline
a & Aa & aa \\ \hline
\end{array} \]

What is the probability of getting \( Aa \)?

**Sum Rule:**
If an event can occur more than one way, the probability of that event is the sum of the independent probabilities.

Use **Sum Rule**:
\[
\left( \frac{1}{2} \times \frac{1}{2} \right) + \left( \frac{1}{2} \times \frac{1}{2} \right) = \frac{1}{2}
\]

(or)
This is a ratio of genotype

What would a ratio of phenotype look like?

Depends on specific alleles we are talking about:

1. dominant and recessive alleles: 
   
   \[
   Aa = AA
   \]
   
   (___phenotypes)

2. codominant alleles
   
   \[
   aa \neq Aa \neq AA
   \]
   
   (___phenotypes)

3. incomplete dominance:
   
   \[
   aa < Aa < AA
   \]
   
   (___phenotypes)
Most human traits are caused by multiple genes rather than a single gene
However there are 1000s of known human traits (most are rare diseases) that are simple: one gene (= “Mendelian Trait”)

Sickle Cell Anemia
Albinism
Cystic Fibrosis
Hemophelia
Polydactyl
Tay Sachs Disease
Achondroplasia
Huntington’s Disease
Phenylketonuria (PKU)
Adenosine deaminase deficiency (ADA)
Consider an example:
Sickle Cell Anemia:
caused by one recessive allele of the β globin gene

\[ \text{aa} = \text{sc anemia} \]
\[ \text{Aa} = \text{healthy (but called “carrier”)} \]
\[ \text{AA} = \text{healthy} \]

\[ \text{Aa} \times \text{Aa} \]
What is the probability of this couple having a child with sc?
What is the probability of having a healthy child?
What is the probability that their healthy child is a carrier?

\[ \text{Aa} \times \text{AA} \]
answer same questions as above
Follow TWO sets of chromosomes through meiosis (note random assortment at the metaphase plate)

maternal
\[ \begin{align*}
A & B \\
A & a
\end{align*} \]

paternal
\[ \begin{align*}
b & b \\
a & a
\end{align*} \]

What is genotype of parent? List the genotypes of the different gametes that this parent can produce. Consider a cross between this individual and another of the same genotype: Use a Punnett square to figure out the probabilities of the various genotypes that can be generated