Modes of Inheritance

I

II

III

IV

Criteria:
1. The trait characteristically appears only in sibs, not in their parents, offspring or other relatives.
2. On the average, one-fourth of the sibs of the propositus are affected.
3. The parents of the affected child may be consanguineous.
4. Males and females are equally likely to be affected.

Autosomal Dominant Inheritance

1. The trait appears in every generation, with no "skipping".
2. The trait is transmitted by an affected person to half of his (her) children.
3. Unaffected persons do not transmit the trait to their children.
4. The occurrence and transmission of trait is not influenced by sex.

Code:
- male
- female
- affected
- carrier (heterozygous)
- consanguineous
- propositus
- mating
- mating

Stereotype pedigree for autosomal dominant inheritance

Stereotype pedigree
Sex-linked Recessive

Criteria:
1. The incidence of the trait is much higher in males than in females.
2. The trait is passed on from an affected man through all his daughters to half of their sons.
3. The trait is never transmitted directly from father to son.
4. The trait may be transmitted through a series of carrier females, and if so, the affected males in a kindred are related to one another through females.

Sex-linked Dominant Inheritance

Criteria:
1. Affected males transmit the trait to all their daughters, none of their sons.
2. Affected females who are heterozygous transmit the condition to half of their children of either sex.
3. Affected females who are homozygous transmit the condition to all their children.
4. Transmission by females follows the same pattern as shown by autosomal dominant inheritance. Therefore sex-linked dominant inheritance can only be distinguished from autosomal dominant inheritance by the progeny of affected males.
Mendelian Inheritance

particulate (discrete) bodies = genes

Gregor Mendel 1865

gene = basic unit of inheritance (segment of DNA)
allele = alternative expression of gene (e.g., A, B, O)
locus = gene’s position on chromosome
genotype = genetic constitution or allelic pair
phenotype = observable or testable characteristics

heterozygous - allelic pair differ (e.g., AO, BO, AB)
homozygous - allelic pair identical (e.g., AA, BB, OO)
dominance - allele expressed in homo- or heterozygous state (e.g., AO, AA, BO, BB)
recessiveness - allele expressed only in homozygous state (e.g., OO)
co-dominance - alleles equally expressed in phenotype (e.g., AB)

Mendel’s Laws:
1. Law of segregation
2. Law of independent assortment

Mendel’s pea plant experiments
- color of endosperm (seed): yellow or green
- stature: short or tall
- shape of ripe seed: smooth or wrinkled
- pod color: green or yellow
- position of blossom: axial or terminal
- etc.

1. observed each plant separately
2. kept generations separate
3. quantified results

<table>
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<th></th>
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<th>Short</th>
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<tr>
<td>Tall</td>
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<tr>
<td></td>
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|       | 3    | 1     | F₂
dominant = Tall  
recessive = Short

T = dominant allele for round seed  
t = recessive allele for wrinkled seed

\[
\begin{array}{ccc}
TT & x & tt \\
\downarrow & & \\
Tt & x & Tt \\
\downarrow & & \\
TT & Tt & tt \\
1 & 1 & 2 & 1 \text{ sort}
\end{array}
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\[
F_1
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t & t \\
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T & Tt & Tt \\
\end{array}
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\[
F_2
\]

\[
\begin{array}{ccc}
T & t \\
T & TT & Tt \\
t & Tt & tt \\
\end{array}
\]

TT or Tt = Tall  
tt = Short

2nd Law of Inheritance (Independent Assortment)

\[
\begin{array}{ccc}
TTRR & x & ttrr \\
\downarrow & & \\
TtRr & x & TtRr \text{ (all yellow & tall)} \\
\downarrow & & \\
Tt & 3 & Tt & 3 & 1
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T = Tall
t = Short

Y = Yellow
y = Green

F₁

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F₂

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Human Inheritance patterns

- dominant, recessive allele
- co-dominant alleles
- pedigree analysis

1. autosomal dominant (e.g., brachydactyly)
2. autosomal recessive (e.g., albinism)
3. sex-linked recessive (e.g., hemophilia)
4. sex-linked dominant (e.g., G-6PD)

ABO blood group

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<thead>
<tr>
<th>Alleles</th>
<th>Genotypes</th>
<th>Phenotypes</th>
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