Mendelian Genetics

- Mendel & the Quantitative approach
- Monohybrid Cross--Law of Segregation
- Dihybrid Cross--Law of Independent Assortment
- Segregation, Independent Assortment & the movement of Chromosomes
- Mendelian Patterns in Human Genetics
Mendel & the Quantitative approach

- Blending inheritance
- Used common garden pea (*Pisum sativum*)
- Short generation time
- Easy to manipulate
- Purebred lines for one or two traits
Technique
Monohybrid Cross

- Purebred lines for one trait
- Studied 7 characters
- P = Parental
- F1/F2 = Filial generation
- Did F3 generation
- Law of segregation
Genetic Symbols

- Genotype
- Phenotype
- Homozygous, heterozygous
- Genes vs alleles
Monohybrid Cross: Mendel’s Postulates

- Unit factors in pairs in individuals
- Two unlike factors are present; one is Dominant & the other is recessive:
- Segregation: each gamete receives one or the other factor
Law of segregation

• During gamete formation, the two members of a gene pair segregate, randomly so that each gamete receives one or the other factor with equal probability.
Test Cross

- Use Homozygous recessive parent
- Sometimes called backcross
- Know what alleles are brought to the cross
Dihybrid Cross--Law of Independent Assortment

- Purebred lines for two characters
- Looked at them independently
- Two 3:1 ratios
- Gives 9:3:3:1 ratio in F2 cross of heterozygous individuals
Dihybrid cross

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Law of Independent Assortment

• During gamete formation, segregation of alleles of one gene is independent of the segregation of another pair of alleles of another gene.

• No mention of chromosomes
• Did not know about meiosis
Segregation, Independent assortment & the movement of chromosomes

- Segregation: separation of homologous chromosome during anaphase I
- Independent Assortment: Similar to random alignment of homologous chromosomes in metaphase I
Math Methods

- Forked line method
- Using phenotypes
- Independently assorting genes
Mendel’s traits

- 7 characters on different chromosomes
- Some were on same chromosome
- Far enough part on chromosome to show independent assortment
Mendelian Patterns in Human Genetics

**Table 3.3 Representative Recessive and Dominant Human Traits**

<table>
<thead>
<tr>
<th>Recessive Traits</th>
<th>Dominant Traits</th>
</tr>
</thead>
<tbody>
<tr>
<td>Albinism</td>
<td>Achondroplasia</td>
</tr>
<tr>
<td>Alkaptonuria</td>
<td>Brachydactyly</td>
</tr>
<tr>
<td>Ataxia telangiectasia</td>
<td>Congenital stationary night blindness</td>
</tr>
<tr>
<td>Color blindness</td>
<td>Ehlers-Danlos syndrome</td>
</tr>
<tr>
<td>Cystic fibrosis</td>
<td>Facioscapulohumeral muscular dystrophy</td>
</tr>
<tr>
<td>Duchenne muscular dystrophy</td>
<td>Huntington disease</td>
</tr>
<tr>
<td>Galactosemia</td>
<td>Hypercholesterolemia</td>
</tr>
<tr>
<td>Hemophilia</td>
<td>Marfan syndrome</td>
</tr>
<tr>
<td>Lesch-Nyhan syndrome</td>
<td>Neurofibromatosis</td>
</tr>
<tr>
<td>Phenylketonuria</td>
<td>Phenylthiocarbamide tasting (PTC)</td>
</tr>
<tr>
<td>Sickle-cell anemia</td>
<td>Porphyria</td>
</tr>
<tr>
<td>Tay-Sachs disease</td>
<td>Widow’s peak</td>
</tr>
</tbody>
</table>

**Diagram:**

- Male
- Female
- Mating
- Parents and 1 boy, 1 girl (in order of birth)
- Dizygotic (two-egg) twins
- Monozygotic (one-egg) twins
- Sex unspecified
- Number of children of sex indicated
- Affected individuals
- Heterozygotes for autosomal recessive gene
- Carrier of sex-linked recessive gene
- Death
- Abortion or stillbirth (sex unspecified)
- Propositus
- Method of identifying persons in a pedigree; here, the propotitus is child 2 in generation II, or II-2
- Consanguineous marriage (marriage of blood relatives)

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## Traits in Humans

<table>
<thead>
<tr>
<th>Disease</th>
<th>Effect</th>
<th>Incidence of Disease</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Caused by Recessive Allele</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Thalassemia (chromosome 16 or 11)</td>
<td>Reduced amounts of hemoglobin; anemia, bone and spleen enlargement</td>
<td>1/10 in parts of Italy</td>
</tr>
<tr>
<td>Sickle-cell disease (chromosome 11)</td>
<td>Abnormal hemoglobin; sickle-shaped red cells, anemia, blocked circulation; increased resistance to malaria</td>
<td>1/625 African Americans</td>
</tr>
<tr>
<td>Cystic fibrosis (chromosome 7)</td>
<td>Defective cell membrane protein; excessive mucus production, digestive and respiratory failure</td>
<td>1/2000 Caucasians</td>
</tr>
<tr>
<td>Tay-Sachs disease (chromosome 15)</td>
<td>Missing enzyme; buildup of fatty deposit in brain; buildup destroys mental development</td>
<td>1/3000 Eastern European Jews</td>
</tr>
<tr>
<td>Phenylketonuria (PKU) (chromosome 12)</td>
<td>Missing enzyme; mental deficiency</td>
<td>1/10,000 Caucasians</td>
</tr>
<tr>
<td>Albinism (chromosome 11)</td>
<td>Missing enzyme; unpigmented skin, hair, and eyes</td>
<td>1/10,000 in Northern Ireland</td>
</tr>
<tr>
<td><strong>Caused by Dominant Allele</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Hypercholesterolemia (chromosome 19)</td>
<td>Missing protein that removes cholesterol from the blood; heart attack by age 50</td>
<td>1/122 French Canadians</td>
</tr>
<tr>
<td>Huntington disease (chromosome 4)</td>
<td>Progressive mental and neurological damage; neurologic disorders by ages 40–70</td>
<td>1/25,000 Caucasians</td>
</tr>
</tbody>
</table>
Dominant Trait: Brachydactyly
Autosomal Dominant

1. Every affected person in the pedigree must have at least one affected parent.
2. Approximately equal numbers of males and females in the pedigree must express the trait, and those individuals should transmit the mutant gene to their progeny.
3. The pedigree must have male and female individuals in each generation who express the mutant gene. In other words, the trait must not skip generations, nor should it appear in males only in one generation, in females only in the next generation, in males only in the generation after that, and so on. That transmission pattern is characteristic of sex-linked inheritance. Father-to-son and mother-to-daughter transmission is expected to occur as frequently as father-to-daughter and mother-to-son transmission in autosomal dominant inheritance.
4. An affected heterozygous individual must transmit the mutant gene to half his or her progeny. Suppose the dominant mutant allele is designated \( A \), and its wild-type allele is \( A^+ \). Then most crosses will be \( A/A^+ \times A^+/A^+ \). From basic Mendelian principles half the progeny will be \( A^+/A^+ \) and the other half will be \( A/A^+ \), showing the trait.
Autosomal Recessive
Pedigree Analysis

(a) Autosomal Recessive Trait

Either I-3 or I-4 must be heterozygous
Recessive traits typically skip generations
Recessive autosomal traits appear equally in both sexes

(b) Autosomal Dominant Trait

I-1 is heterozygous for a dominant allele
Dominant traits seldom skip generations
Affected individuals all have an affected parent. Dominant autosomal traits appear equally in both sexes.