Foundations of Genetics
Mendelism

Patterns of Inheritance
Heredity - Genetics

• Heredity: is the passage of characters from parents to offspring.
• Genetics: is the study of heredity.
• John Gregor Mendel was the first to scientifically study inheritance. Fig 11.1
• He established the fundamental laws of heredity by studying pea plants. Fig 11.2
• He kept detailed records of his experiments.
• He made hypotheses and tested them with a flawless scientific method. He used statistical analysis. Fig 11.3
Basic Terms

• **Characters/traits** are controlled by discrete units called *genes*. For example, Height of Pea plant is a trait and so are the color of flower, seed or pod. Table 11.1

• A **gene** has 2 alternate versions called **Alleles**.

• Parents carry 2 alleles for each character but the gametes carry only 1 allele. In body cells with 2 alleles when both are similar = **homozygous** (YY or yy) and when different = **heterozygous** (Yy).

• When 2 different alleles come together only one determines the appearance of body (dominant) and the other remains hidden (recessive). For example Yellow seed color dominates green seed color. Though recessive allele does not express but maintains its identity.

• **Phenotype** = appearance, yellow seed  **Genotype** = genetic make-up, Yy
Probability and Mendel

• If we toss a coin it can land either heads or tails up. There are total events possible, 2 in this example. If we say what is the possibility of heads landing up? It is ½ or 50%.

• **Probability** is the possibility of occurrence of an event out of the total possible events. Fig 11.8

• Mendel used probability to predict the outcome of testing his hypotheses on inheritance of characters in Pea plant.

• He studied 7 characters. Table 11.1

• For example Flower Color. 2 alleles for this gene control Purple and white color.

• Purple expresses in heterozygous condition. It means Purple is dominant over white color allele.

• Allele for Purple color = P and allele for white color = p
Mendel’s 4 Hypotheses

• Mendel’s 4 hypotheses: 1. 2 alternate forms of each gene called **alleles**.

• 2. An organism inherits 2 alleles, 1 from each parent. When similar like PP or pp called **Homozygous** and when different like Pp called **Heterozygous**.

• 3. If 2 alleles are different, one determines the appearance of organism = **dominant** and other does not contribute to appearance = **recessive**.

• 4. An egg or sperm carries only 1 allele of each pair because these are separated from each other during gamete (meiosis) formation. This is now called **Mendel’s Law of Segregation**.
Monohybrid Cross
an introduction

- Mendel tested inheritance of one character at one time. It is called a Monohybrid Cross. Fig 11.5 / 11.8
- Mendel sowed pea plants and maintained records and kept seeds separate if they yielded only purple flowers or white flowers and called them pure purple and pure white plants.
- Pea plants normally undergo self-fertilization, the pollen grains (male part) come in contact with stigma of carpel (female part) of same flower.
- Cross: Mendel manipulate the Pea flowers. He removed the stamens (produce pollens) of one pure flower, say white, and brought the pollens from a pure purple flower and touched on the stigma of white flower to cause Cross-fertilization.
Law of Segregation

• Law of Segregation: 2 alleles of a gene in an organism separate from each other at the time of gamete formation and gametes fertilize at random.
• Mendel tested the hypothesis with the help of a monohybrid cross.
• P = parents   G = gametes   F₁ = 1ˢᵗ daughter generation
• F₂ = 2ⁿᵈ daughter generation
• We take the example of a monohybrid cross between pure purple (PP) and pure white (pp) flower plants.
• Phenotypic Ratio is based on appearance of flowers = 3 : 1
• Genotypic Ratio is based on alleles present = 1 : 2 : 1
Law of Segregation
A Punnet Square

P
PURE PURPLE
PP
G
P
X
F1
Pp (all purple)

F2

PP
Purple

pp
Purple

pp
White

Phenotypic Ratio
Purple : White
3 : 1

Genotypic Ratio
PP : Pp : pp
1 : 2 : 1
Dihybrid Cross

When Mendel studied 2 characters at same time he called it a Dihybrid Cross.

- yellow-round   yellow-wrinkled   green-round   green-wrinkled
- 9               :             3               :           3           :          1
Incomplete Dominance

- Fig 11.14
- In Japanese 4 O’Clock, a cross between red and white flowers gives pink flowers in F1.
- When allowed to self-fertilize the pink flowered plants gives a ratio of Red 1 : Pink 2 : White 1 (1:2:1). Therefore the phenotypic ratio and genotypic ratio are same.
Polygenic Inheritance

• In humans height, skin color and body weight are not controlled by single genes. Therefore these characters do not obey Mendel’s laws.

• These exhibit continuous variation and when marriage between a black and white person, F1 individual resembles neither of the Parents. Same thing happens for human height. Fig 11.12
Multiple Alleles

- Inheritance of ABO blood groups in humans - Fig 11.19
- 3 alleles, IA, IB and i, control inheritance of 4 blood groups A, B, AB and O.
- Blood group A has antigen A and antibody b.
- Blood group B has antigen B and antibody a.
- Blood group AB has antigens A and B but no antibodies.
- Blood group O has no antigens but has antibodies a and b.
- Therefore blood group AB is universal recipient and blood group O is universal donor.
- Any of these 4 blood group can be Rh+ or Rh-. Rh is taken from the Rhesus monkey. Most of human blood agglutinate with Rhesus factor in its blood and are called +. Rh+ is dominant over Rh-.
- Rh- mother cannot bear a Rh+ fetus 2nd time onwards due to lot of antibodies in her blood formed during 1st pregnancy. It is called Rh incompatibility.
## ABO Blood Groups

- ABO blood groups

<table>
<thead>
<tr>
<th>Blood Group</th>
<th>Antigen</th>
<th>Antibody</th>
<th>Genotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>A</td>
<td>A</td>
<td>b</td>
<td>$I^A I^A, I^A i$</td>
</tr>
<tr>
<td>B</td>
<td>B</td>
<td>a</td>
<td>$I^B I^B, I^B i$</td>
</tr>
<tr>
<td>AB</td>
<td>A, B</td>
<td>None</td>
<td>$I^A I^B$</td>
</tr>
<tr>
<td>O</td>
<td>None</td>
<td>Both a, b</td>
<td>ii</td>
</tr>
</tbody>
</table>
Non-disjunction

- Fig 11.23
- During gamete formation sometimes the homologous chromosomes fail to separate during anaphase-1 or anaphase-2. It leads to formation of gametes with n-1 or n+1 chromosomes. So in humans instead of normal 23 chromosomes some gamete may have 22 or 24 chromosomes. On fertilizing a normal gamete they produce individuals with 45 or 47 chromosomes. These are called syndromes.
- Down’s syndrome is the most common type and have 3 chromosomes of 21st chromosome instead of 2. Fig 11.24
Human Genome

• The genome is the full complement of genetic information of an organism (i.e., all of its genes and other DNA)

• Human genome has 20000-250000 genes represented by only 1% of total human DNA.
Genetic Engineering

A gene transfer experiment occurs in four stages

1. **Cleaving DNA**
   - cutting the source and vector DNA using Restriction Enzymes.

2. **Producing recombinant DNA**
   - Sealing source and vector DNA using DNA Ligase to form Recombinant DNA and then transferring the DNA into the target cells

3. **Cloning**
   - introducing DNA-bearing vectors into target cells and then allowing the target cells to reproduce

4. **Screening**
   - selecting the particular infected cells that have received the gene of interest
Embryonic Stem Cells

- **Embryonic stem cells** are special cells that form early in development and each has the capacity to develop into a healthy individual.

- **Totipotent** is the ability of cells, such as stem cells, to have the ability to form any body tissue, and even an adult animal.