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 $\frac{1}{2}$ 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                        PURE WHITE
PP                                          pp
G                      P                                            p
X
F1                                           Pp (all purple)

F_2
male
female

Phenotypic Ratio
Purple  White
3    :    1

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

- When Mendel studied 2 characters at the 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, I^A, I^B 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, I^A_i)</td>
</tr>
<tr>
<td>B</td>
<td>B</td>
<td>a</td>
<td>(I^B_i, I^B_i)</td>
</tr>
<tr>
<td>AB</td>
<td>A, B</td>
<td>None</td>
<td>(I^A_iB)</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
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

2. **Cleaving DNA**
   - cutting the source and vector DNA

3. **Producing recombinant DNA**
   - placing the DNA fragments into vectors and then transferring the DNA into the target cells

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

5. **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.