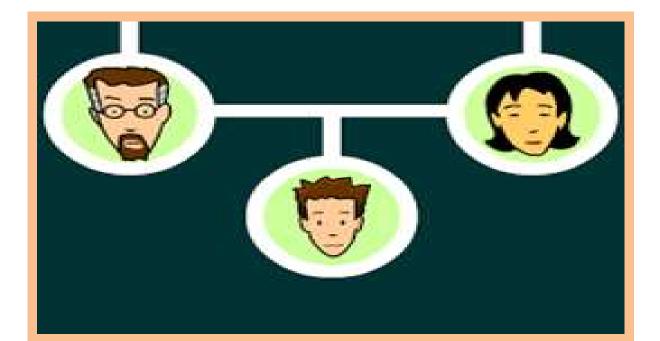
## GCOE

# CLASS : B. Ed Sem.2nd SUBJECT: Pedagogy of Bio-Science INCHARGE: Dr. JYOTI PARIHAR

# UNIT :5 SUB UNIT :5.2

Mendelian inheritance and Deviations from Mendelism-Incomplete dominance, co-dominance, multiple alleles and inheritance of blood groups.



## PRINCIPLES OF INHERITANCE AND VARIATION

## **IMPORTANT TERMS**

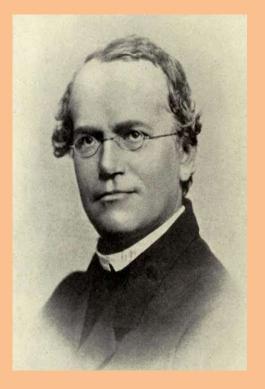
- Genetics:- Branch of life science dealing with study of heredity and variation.
- □ <u>Heredity:-</u> Transmission of characters from parents to their offsprings.
- **Variation:-** Difference among offsprings and with their parents.
- **Hereditary variation:-** Genetic and inheritable.
- **Environmental variation:-** Acquired and non-heritable.
- □ <u>Inheritance:-</u> Reception of genetic qualities by transmission from parents to offsprings.

- Gene:- Unit of inheritance.
- **<u>Trait:-</u>** Features of an individual.
- □ <u>Hybrid:</u> Progeny obtained by crossing two parents that differ in characters.
- **Gene Locus:-** Portion of chromosome representing a single gene.
- □ <u>Homozygous condition:-</u> When both the genes responsible for a particular trait are identical. For e.g., T---T, t---t condition in case of height.
- □ <u>Heterozygous condition:-</u> When both the genes of a character are unidentical. For e.g., T---t condition.

- □ <u>Phenotypic Ratio:</u> External manifestation of gene products brought to expression. It can be known through direct observation. It may change with time and environment.
- □ <u>Genotypic Ratio:</u> External and internal manifestation of gene products in an organism. It remains the same throughout the life.
- □ <u>F1 Generation:-</u> Offsprings produced by crossing two genetically different parents. First Filial Generation.
- □ <u>F2 Generation:</u> Generation obtained by interbreeding or selfing F1 individuals. Second Filial Generation.
- □ <u>F3 Generation:</u> Offspring arises by selfing F2 individuals. Third Filial Generation.

## **GREGOR JOHANN MENDEL**

- **□** 1822 1884.
- Austrian monk.
- □ Hybridization Experiment with pea plants.
- Published his results "Translation of the characters" in the natural history of society of brunn.
- □ Regarded as the "Father of Genetics".



<u>SEVEN PAIRS OF CONTRASTING</u> CHARACTERS SELECTED BY MENDEL FOR HIS EXPERIMENT							
	Height	Seed Shape	Seed Color	Seed Coat Color	_	Pod Color	Flower Position
Dominant	Tall	Round	O Yellow	Green	) Inflated (full)	Green	Axial
Recessive Trait	Short	Wrinkled	Green	White	Constricted (flat)	Yellow	Terminal

# WHY MENDEL SELECTED PEA PLANT?

- Dure varieties are available.
- Pea plants are easy to cultivate.
- Life cycle of plants are only few months.
- Results can be got early.
- Contrasting traits are observed.
- Flowers are bisexual and normally self-pollinated.
- Flowers can be cross pollinated only manually.
- Hybrids are fertile.

## MENDEL'S PRINCIPLES OF INHERITANCE

□ Principle of Paired Factors.

□ Principle of Dominance.

□ Principle of Segregation.

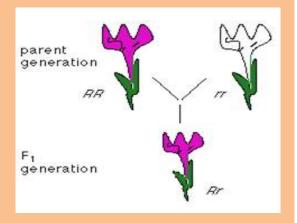
□ Principle of Independent Assortment.

### PRINCIPLE OF PAIRED FACTORS

- A character is represented in an organism by atleast two factors.
- □ The two factors lie on the same homologous chromosomes at the same locus.
- □ They may represent the same (homozygous, e.g., TT in case of pure tall pea plants) or alternate expression (heterozygous, e.g., Tt in case of hybrid tall pea plants) of the same character.

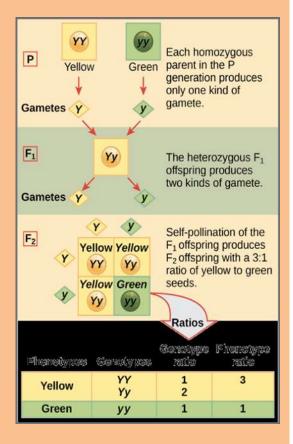
### PRINCIPLE OF DOMINANCE

- Every gene has two alleles that can code for a trait.
- □ One allele is dominant, meaning it will always show.
- Other allele is recessive, meaning it will be masked by the presence of the dominant allele.
- Hybrids will always show the dominant phenotype.



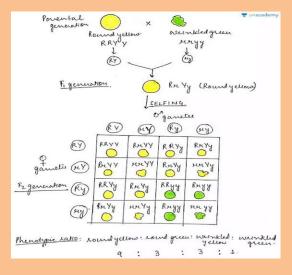
## PRINCIPLE OF SEGREGATION

- □ During gamete (Egg or Sperm) formation, the two alleles of a gene get separated.
- Also called Law of Splitting of Hybrids or Law of Purity of Gametes.
- □ Alleles for a trait then recombine at fertilization producing genotype for the traits of the offsprings.
- E.g., Monohybrid cross.



#### PRINCIPLE OF INDEPENDENT ASSORTMENT

- □ Each character is independently inherited in the next generation irrespective of the source of character in parent generation.
- □ Each gene set is present, not as single thread of DNA but as independent pieces called Chromosome.
- **E**.g., Dihybrid cross.
- Genotypic ratio:- 1:2:1:2:4:2:1:2:1



## REASONS FOR MENDEL'S SUCCESS

- □ Mendel selected only pure breeding varieties of pea.
- □ He studied only those traits, which do not show any gene to gene interaction.
- □ He took one or two characters at a time.
- □ Pea is the ideal plant for cross breeding manually.
- □ He kept a complete accurate record of his breeding experiments.
- □ He used statistical methods and law of probability for analysing his results.
- □ He was lucky in selecting those traits, genes of which were located on different chromosomes.

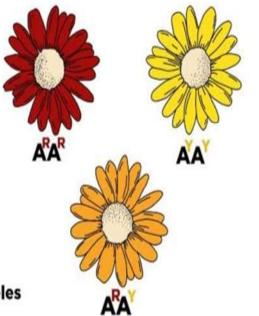
## **MENDEL'S FAILURES**

□ Principle of Incomplete Dominance.

□ Principle of Co-Dominance.

## **INCOM PLETE DOMINANCE**

- □ Neither allele is dominant.
- □ When an organism is heterozygous for a trait, it will show a third phenotype.
- □ In the example, the letter 'A' represents the gene.
- □ 'R' and 'Y' represent the eles different alleles.



### **CO-DOMINANCE**

- □ Co- means together.
- Co-dominant means equal in dominance.
- □ Alleles show approximately equal effect in individuals; alleles are equally detectable in individuals.
- □ Phenotypes for both alleles are exhibited in the heterozygous.
- □ The hybrid shows neither of the parents' trait, instead, a third, different phenotype.
- E.g., Human blood groups.

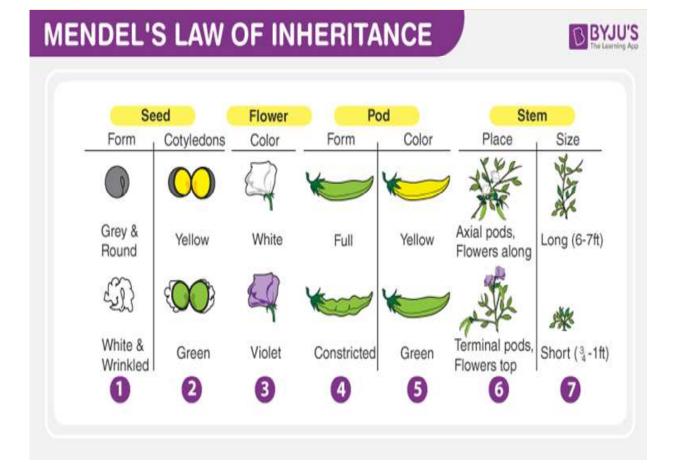
There are 3 different alleles, 6 different genotypes control 4 different type of sugar Phenotypes :

Blood Group	Genotype	Types of Sugar	
A	I <sup>A</sup> I <sup>A</sup> , I <sup>A</sup> i	A	
В	I <sup>B</sup> I <sup>B</sup> , I <sup>B</sup> i	В	
AB	I <sup>A</sup> I <sup>B</sup> ,	both A & B	
0	ii	No sugar alleles	

In humans, blood group AB shows co-dominance as both the alleles I<sup>A</sup> and I<sup>B</sup> express themselves fully in presence of each other.

#### **Mendel's Laws of Inheritance**

- Inheritance can be defined as the process of how a child receives genetic information from the parent. The whole process of heredity is dependent upon inheritance and it is the reason that the offsprings are similar to the parents. This simply means that due to inheritance, the members of the same family possess similar characteristics.
- It was only during the mid 19th century that people started to understand inheritance in a proper way. This understanding of inheritance was made possible by a scientist named Gregor Mendel who formulated certain laws to understand inheritance known as Mendel's laws of inheritance.



Between 1856-1863, Mendel conducted the hybridization experiments on the garden peas. During that period, he chose some distinct characteristics of the peas and conducted several cross-pollination/ artificial pollination on the pea lines that showed stable trait inheritance and underwent continuous selfpollination. Such pea lines are called true-breeding pea lines.

#### Why was Pea Plant Selected for Mendel's Experiments?

- He selected a pea plant for his experiments:
- The pea plant can be easily grown and maintained.
- Ther are naturally self-pollinating but can also be cross-pollinated.
- It is an annual plant, therefore, many generations can be studied within a short period of time.
- It has several contrasting characters.

Mendel conducted two main experiments to determine the laws of inheritance. These experiments were:

- 1. Monohybrid Cross Experiment
- 2. Dihybrid Cross Experiment

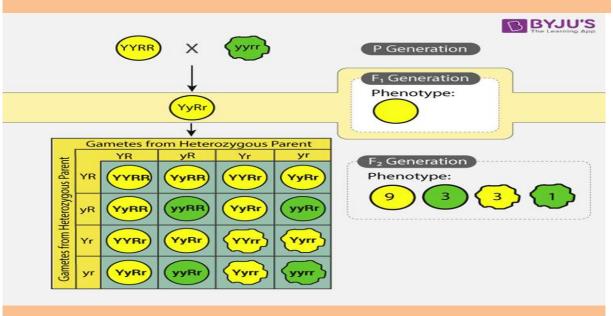
While experimenting, Mendel found that certain factors were always being transferred down to the offsprings in a stable way. Those factors are now called genes i.e. genes can be called as the units of inheritance.

#### **Mendel's Experiments**

 Mendel experimented on a pea plant and considered 7 main contrasting traits in the plants. Then, he conducted both the experiments to determine the aforementioned inheritance laws. A brief explanation of the two experiments is given below.

#### Monohybrid Cross

In this experiment, Mendel took two pea plants of opposite traits (one short and one tall) and crossed them. He found the first generation offsprings were tall and called it F1 progeny. Then he crossed F1 progeny and obtained both tall and short plants in the ratio 3:1. Mendel even conducted this experiment with other contrasting traits like green peas vs yellow peas, round vs wrinkled, etc. In all the cases, he found that results were similar. From this, he formulated the <u>laws of Segregation And</u> <u>Dominance</u>.



After conducting for other traits, the results were found to be similar. From this experiment, Mendel formulated his second law of inheritance i.e law of Independent Assortment.

#### **Mendel's laws**

The two experiments lead to the formulation of Mendel's laws known as laws of inheritance which are:

- Law of Dominance
- Law of Segregation
- Law of Independent Assortment

#### Law of Dominance

This is also called as Mendel's first law of inheritance. According to the law of dominance, hybrid offsprings will only inherit the dominant trait in the phenotype. The alleles that are suppressed are called as the recessive traits while the alleles that determine the trait are known as the dormant traits.

#### Law of Segregation

The law of segregation states that during the production of gametes, two copies of each hereditary factor segregate so that offspring acquire one factor from each parent. In other words, allele (alternative form of the gene) pairs segregate during the formation of gamete and re-unite randomly during fertilization. This is also known as Mendel's third law of inheritance.

#### Law of Independent Assortment

Also known as Mendel's second law of inheritance, the law of independent assortment states that a pair of trait segregates independently from another pair during gamete formation. As the individual <u>heredity</u> factors assort independently, different traits get equal opportunity to occur together.

### **DEVIATIONS FROM MENDELISM**

## Co-dominance , incomplete dominance and multiple alleles

#### 5.2.2.2 Co-dominance

Till now we were discussing crosses where the F, resembled either of the two parents (dominance) or was in-between (incomplete dominance). But, in the case of co-dominance the F, generation resembles both parents. A good example is different types of red blood cells that determine ABO blood grouping in human beings. ABO blood groups are controlled by the gene I. The plasma membrane of the red blood cells has sugar polymers that protrude from its surface and the kind of sugar is controlled by the gene. The gene (I) has three alleles  $I^A$ ,  $I^B$  and i. The alleles  $I^A$  and  $I^B$  produce a slightly different form of the sugar while allele i doesn't produce any sugar. Because humans are diploid organisms, each person possesses any two of the three I gene alleles.  $I^A$  and  $I^B$  are completely dominant over *i*, in other words when  $I^{A}$  and *i* are present only  $I^{A}$  expresses (because *i* does not produce any sugar), and when  $I^B$  and *i* are present  $I^B$  expresses. But when  $I^{A}$  and  $I^{B}$  are present together they both express their own types of sugars: this is because of co-dominance. Hence red blood cells have both A and B types of sugars. Since there are three different alleles, there are six different combinations of these three alleles that are possible a total of six different genotypes of the human ABO blood types (Table 5.2). How many phenotypes are possible?

Allele from Parent 1	Allele from Parent 2	Genotype of offspring	Blood types of offspring
I A	ΙΑ	ΙΑΙΑ	А
ΙΑ	I <sup>B</sup>	$I^A I^B$	AB
I <sup>A</sup>	i	I^ i	А
I <sup>B</sup>	ΙΑ	$I^A I^B$	AB
I <sup>B</sup>	I <sup>B</sup>	I <sup>B</sup> I <sup>B</sup>	В
I <sup>B</sup>	i	I <sup>B</sup> i	В
i	i	i i	0

Table 5.2: Table Showing the Genetic Basis of Blood Groups in Human Population

7

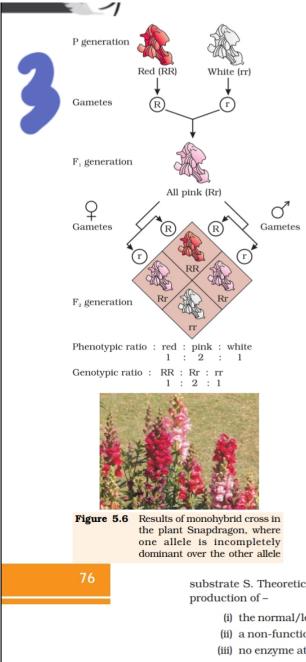
#### BIOLOGY



Do you realise that the example of ABO blood grouping also provides a good example of **multiple alleles**? Here you can see that there are more than two, i.e., three alleles, governing the same character. Since in an individual only two alleles can be present, multiple alleles can be found only when population studies are made.

Occasionally, a single gene product may produce more than one effect. For example, starch synthesis in pea seeds is controlled by one gene. It has two alleles (**B** and **b**). Starch is synthesised effectively by **BB** homozygotes and therefore, large starch grains are produced. In contrast, bb homozygotes have lesser efficiency in starch synthesis and produce smaller starch grains. After maturation of the seeds, BB seeds are round and the bb seeds are wrinkled. Heterozygotes produce round seeds, and so **B** seems to be the dominant allele. But, the starch grains produced are of intermediate size in **Bb** seeds. So if starch grain size is considered as the phenotype, then from this angle, the alleles show incomplete dominance.

Therefore, dominance is not an autonomous feature of a gene or the product that it has information for. It depends as much on the gene product and the production of a particular phenotype from this product as it does on the particular phenotype that we choose to examine, in case more than one phenotype is influenced by the same gene.



#### 5.2.2.1 Incomplete Dominance

When experiments on peas were repeated using other traits in other plants, it was found that sometimes the F, had a phenotype that did not resemble either of the two parents and was in between the two. The inheritance of flower colour in the dog flower (snapdragon or Antirrhinum sp.) is a good example to understand incomplete dominance. In a cross between true-breeding red-flowered (RR) and truebreeding white-flowered plants (rr), the F, (Rr) was pink (Figure 5.6). When the  $F_1$  was self-pollinated the  $F_{2}$  resulted in the following ratio 1 (RR) Red: 2 (Rr) Pink: 1 (rr) White. Here the genotype ratios were exactly as we would expect in any mendelian monohybrid cross, but the phenotype ratios had changed from the 3:1 dominant : recessive ratio. What happened was that R was not completely dominant over r and this made it possible to distinguish Rr as pink from RR (red) and rr (white).

Explanation of the concept of dominance: What exactly is dominance? Why are some alleles dominant and some recessive? To tackle these questions, we must understand what a gene does. Every gene, as you know by now, contains the information to express a particular trait. In a diploid organism, there are two copies of each gene, i.e., as a pair of alleles. Now, these two alleles need not always be identical, as in a heterozygote. One of them may be different due to some changes that it has undergone (about which you will read further on, and in the next chapter) which modifies the information that particular allele contains.

Let's take an example of a gene that contains the information for producing an enzyme. Now there are two copies of this gene, the two allelic forms. Let us assume (as is more common) that the normal allele produces the normal enzyme that is needed for the transformation of a

substrate S. Theoretically, the modified allele could be responsible for

- (i) the normal/less efficient enzyme, or (ii) a non-functional enzyme, or
- (iii) no enzyme at all

PRINCIPLES OF INHERITANCE AND VARIATION



In the first case, the modified allele is equivalent to the unmodified allele, i.e., it will produce the same phenotype/trait, i.e., result in the transformation of substrate S. Such equivalent allele pairs are very common. But, if the allele produces a non-functional enzyme or no enzyme, the phenotype may be effected. The phenotype/trait will only be dependent on the functioning of the unmodified allele. The unmodified (functioning) allele, which represents the original phenotype is the dominant allele and the modified allele is generally the recessive allele. Hence, in the example above the recessive trait is seen due to non-functional enzyme or because no enzyme is produced.

### **INCOMPLETE DOMINANCE**



In incomplete dominance, genes are not related as dominant and recessive but express themselves when presenting together in hybrid.

## 10

Hence,  $oldsymbol{F_1}$  hybrid exhibit character intermediate to the effect of two parental alleles.

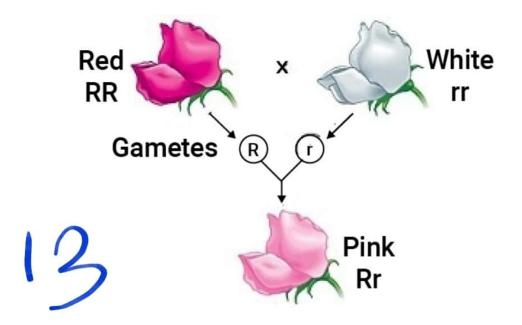
# 11

Hence, here a third character is also seen in addition to two characters that are present in the presence of homozygous conditions or non-hybrid conditions of alleles.

# 12

EXAMPLE

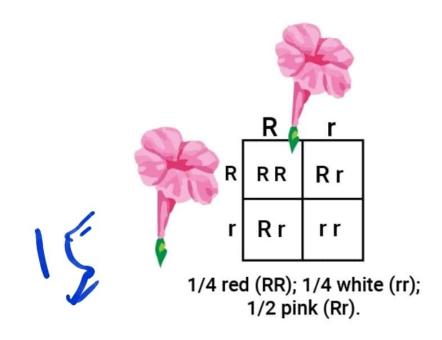
Inheritance of flower color in Four'o clock (Mirabilis jalapa) and Snapdragon (Dog flower or Antirrhinum) are a good example of Incomplete dominance.



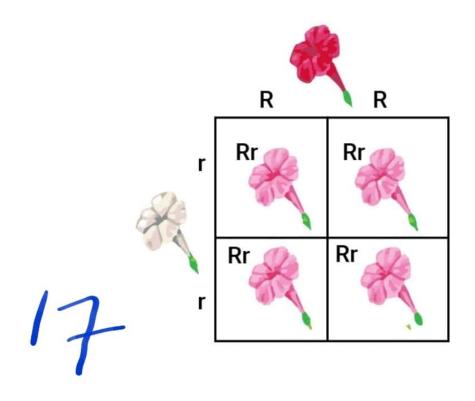
In these plants, a cross between true breeding red-flowered plants (RR) and true breeding white flowers (rr) produce  $F_1$  offspring with pink colour.



## Hence, here an intermediate character (pink) is seen which is a hybrid of both red and white colour.



When  $F_1$  Pink flowered plants are self-pollinated or crossed among themselves,  $F_2$  plants with red (RR), pink (Rr) and white (rr) appeared in ratio 1:2:1.



In  $F_1$ , 'R' ( red color) was not completely dominant over 'r' (white color), so 'Rr' appeared as pink which is an intermediate expression of two alleles.

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Here, Phenotypic ratio is same as the genotypic ratio (1:2:1) of a monohybrid cross, but this ratio is changed from Mendelian 3 (dominant): 1 (recessive) ratio.

### **MULTIPLE ALLELES**

FIGURE 7.12: A dihybrid cross explaining epistasis (see text).

#### 7.6.5. MULTIPLE ALLELES

Simultaneous occurance of more than two alternative forms (alleles) of a gene within of species population at a given gene locus on a chromosome or its homologue are known as multiple alleles.

The conclusion drawn from Mendel's experiments of inheritance provide the concept that each gene has two alternative forms or allelomorphs (one is dominant and the other is recessive) located on the same gene locus. Out of a pair of alleles, one is wide and the other is mutant. It is, therefore, expected that if a wild form of gene can mutate to produce one mutant, it can also produce two or more than two mutants. Therefore, a gene locus can have more than two allelomorphs.

Existence of multiple alleles has been demonstrated in — (i) Colour loci in corns ; (ii) Skin colour in rodents ; (iii) ABO blood groups in humans ; (iv) Eye colour in *Drosophila* ; and many other organisms. An example of multiple alleles in colour loci in corns is given below :

The corn varieties have been found to possess several series of multiple alleles. One such series is located at R locus on chromosome 10. The R locus is responsible for controlling anthocyanin pigment (a phenolic compound) of seed and plant. This locus possesses 4 alleles earlier designated as  $- R^r$ ,  $R^g$ ,  $r^r$ ,  $r^g$ , These alleles have now been renamed as SP,  $S_P$ , sP and sp due to their duel functions. The functions of these alleles are —

SP = Coloured seeds and red coloured plant

Sp = Coloured seeds and green coloured plant

sP = Colourless seeds and red coloured plant

sp = Colourless seeds and green coloured plant

## Inheritance of blood groups

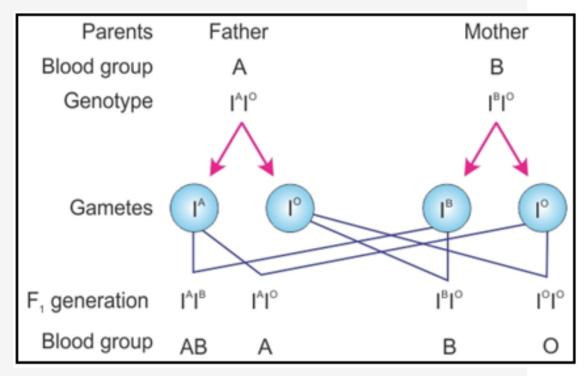
Blood groups are inherited from our biological parents in the same way as eye colour and other genetic traits.

Within the ABO Blood Group system, the A and B genes are co-dominant, i.e. these will be expressed whenever the gene is present. The O gene is silent and only expressed when neither A nor B is present.

The Rh(D) group is also dominant and will be expressed if inherited from either parent. If the Rh(D) gene is not inherited from either biological parent then the individual will be Rh(D)negative.

The following charts outline the possible ABO and Rh(D) group combinations of parents and the possible blood types of their biological children.

Given, the father has blood group A, the mother has blood group B and the child has blood group O. O is a recessive blood group. Therefore, the child will have homozygous recessive alleles  $I^{O}I^{O}$ . This is possible only when both his parents are heterozygous and has allele  $I^{O}$  as one of their alleles. Hence, the genotype of the father will be  $I^{A}I^{O}$  and that of the mother will be  $I^{B}I^{O}$ .



The possible blood groups in the offspring of this couple are AB, A, B and O.

## THANKS