Bioengineering: An Interface with Biology and Medicine
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Lecture – 18
Genetics-III

Welcome to MOOC-NPTEL course on bioengineering, an interface with biology and medicine. Today, we will discuss some deviations from Mendelian genetics where you will understand that not everything obeys Mendel's law.

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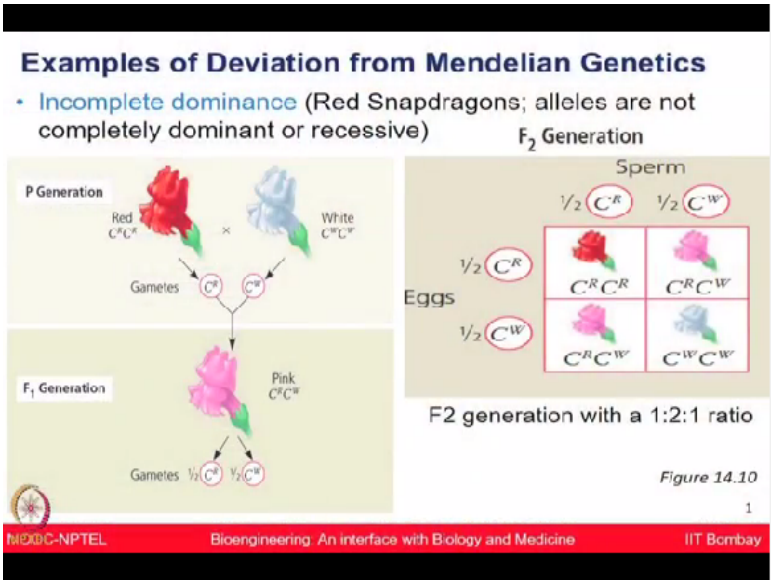

So we have seen in Mendel case, you know, it was purple versus white and always only purple in the F1 generation and then they segregate again in a purple versus white. But there are examples like a flower which is a red snapdragon.

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That flower if you do a cross, so in this case you have, these are the parental gametes. One is a red colour flower, one is white colour flower and on doing the cross from these gametes, it is forming RW which is neither red nor white and it shows the pink colour gametes in the hybrid and now when they do the cross from this, so you got 1 red, 1 white and 2 pink, alright. So this is not following the Mendelian law of dominance, right.

It is not showing you that 3:1 pattern. At the phenotype level, we have ratio which is 1:2:1. In the Mendelian case when we saw the law of segregation, where you observed this number 1:2:1 at the genotype level, right. When these numbers are shown where these ratios denote genotypic ratio in which context or where it can be phenotypic ratio. So for example in the case of incomplete dominance, it is phenotypic ratio which is 1:2:1.

If you have done the cross of the red flower versus white flower, the gametes which were produced from the F1 generation of the pink flower, you did again a cross of that in the F2 generation, you have again sperm and eggs from these 2, RWRW, you are doing cross, 1 red, 1 white and 2 pink flowers were generated. The phenotypic ratio here becomes 1:2:1.

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Another characteristic which is very interesting is albinism where there is some deficiency of the colours which gives you the black shade. And those individuals are having white hair and no pigmentation is one of the lacking characteristics in those individuals which is derived from some genes which are recessively inherited and they are going to only show up in the homozygous individuals.

So those individuals who are carrying it both, aa, only they are going to show you the albinism property. So let us take and please do a cross yourself now.

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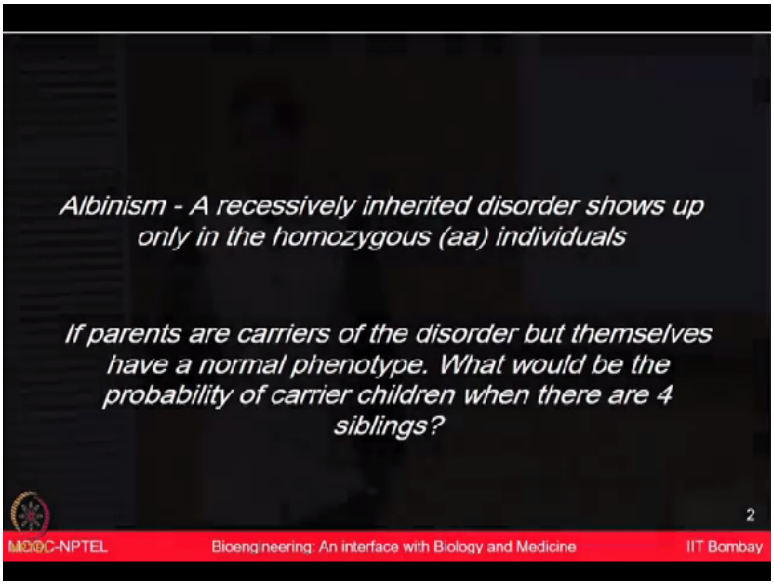

If there are parents who look normal but they are the carriers of this albinism property. So

parents are looking normal but they might be carrying the alleles of this property of albinism, the disorder.

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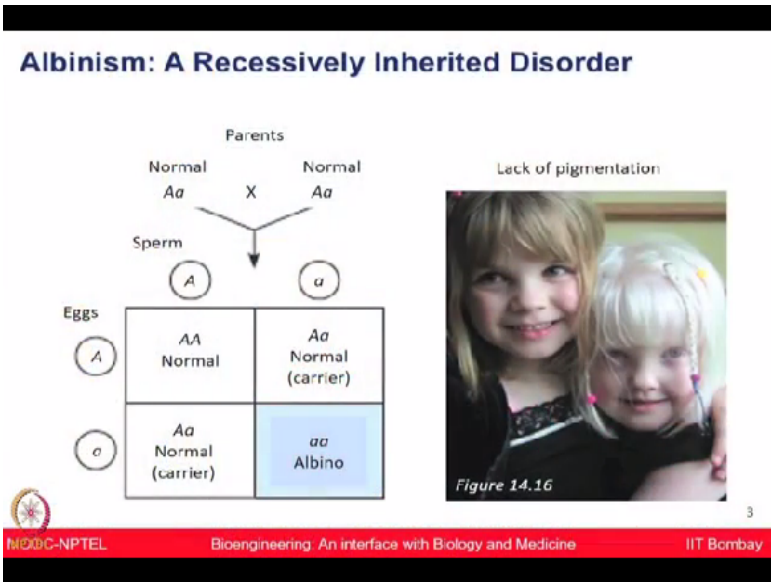

So they are normal phenotype. What would be the probability of their children as a carrier for this particular disease as well as how many of them will be totally albino. I think genetics questions are always good idea that, you know, things look very simple but do it. Just make the cross and think about what should be genotype? How to denote the gametes and then the cross will become very simple.

"Professor - student conversation starts" Both the parents are carriers of the disease? Both the parents are carrier. **"Professor - student conversation ends."** So it means the parents genotype will be what? Heterozygous it will be. So they are looking normal in appearance but the heterozygous, they are carrier and when there is a 1 recessive character, it is not going to show until unless it becomes homozygous recessive.

So now when you do the cross of the 4 child from the family, there is a possibility only 1 of them will become albino and 2 will be actually carrier. So 3 of them are looking normal but of those 3, 2 will be the carrier who will carry the genes again to the next generation. This is how one could study these things in a given family and then try to make some sort of pedigree analysis that what could be the pattern of that. And you see these kind of, you know, pattern of albino child that,

you know, who are looking white, lack of pigmentation in these childs.

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Let us think about some more complex properties and something which is much more related to all of us. Think about the blood, blood groups. I am sure everybody would have, you know, gone to some clinic to know that what their blood groups are, right. These blood groups are derived from the same gene but multiple alleles.

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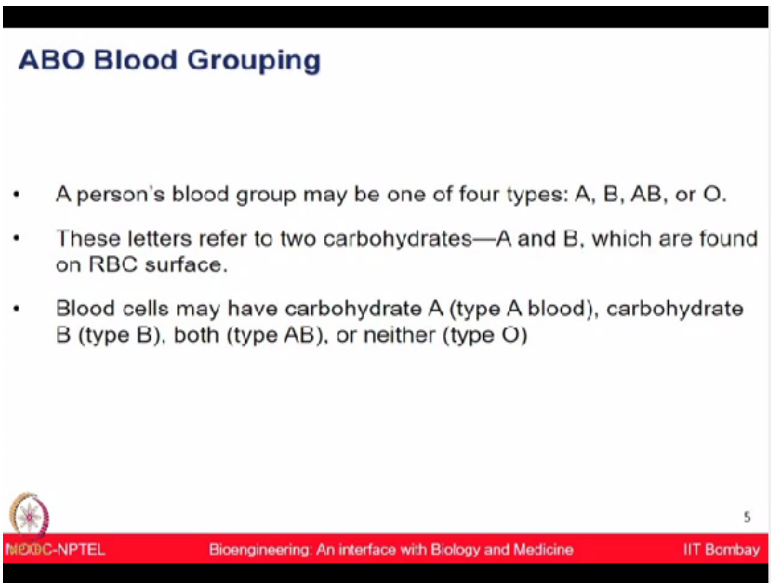

And these alleles, so let us say, you know, I am sure you are aware of there are 4 different type of blood groups, A, B, AB, or O, right. And these nomenclature, these names comes from some sugar, some carbohydrates which are present on the RBCs.

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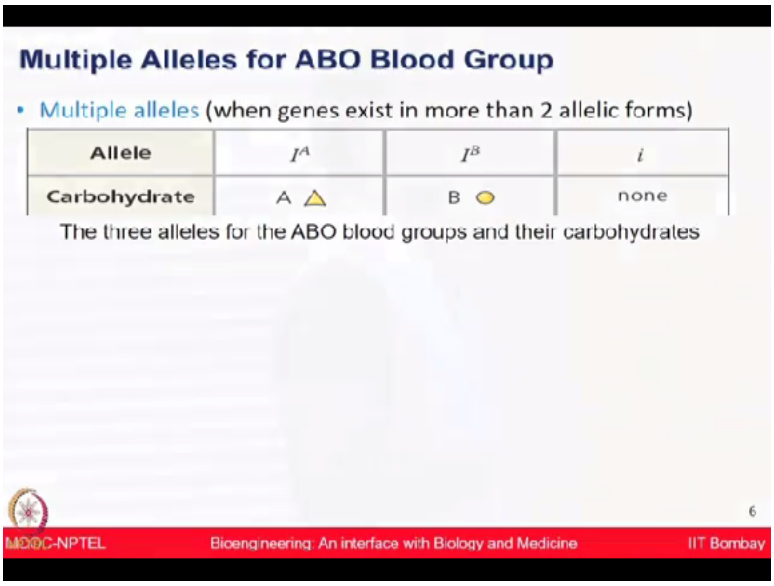

So on the red blood cells, sometimes this carbohydrate A is present and that type of blood is known as blood group A or carbohydrate B is present that is known as blood group B or both of them are present that is known as AB or if both of them are absent, that is known as O type. So let us kind of, you know, start putting this in genetic terms. So we are saying that A and B are dominant traits in this case, right.

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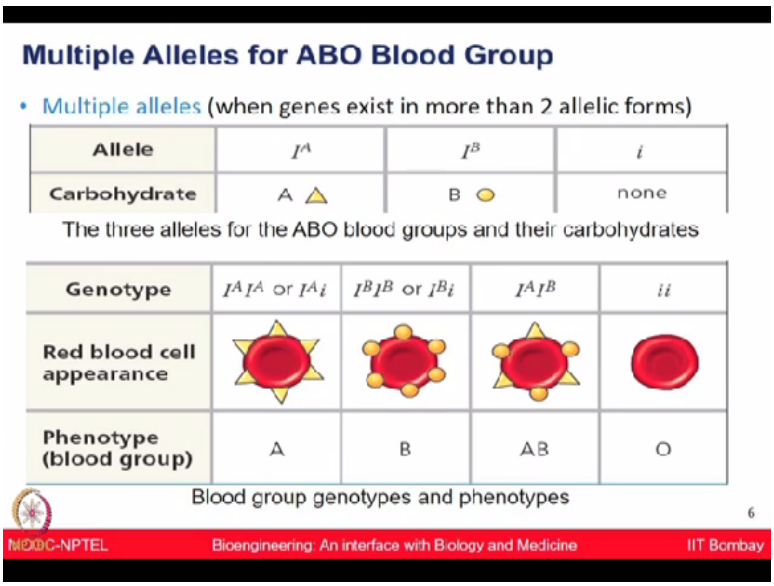

We are talking of multiple alleles of a given gene. So for A blood group, IA is how you are denoting the A blood group. B blood group, IB. When it is a small form of allelic form, you are saying it is i which is none of the carbohydrate is present. In this case, A carbohydrate is present

in the A blood group and B carbohydrate present in the B blood group. Now if you go further, you can start thinking about 2 possibilities over there when we think about blood grouping or any dominance law for that matter.

Even if we have IA and i, that will still appear a dominant trait or just think about the same logic what we thought for purple and white colours, right. Or you have IA and IA, that is also dominant and is also going to show you the A blood type trait. In the B blood group, we have IBIB or IB and a i. In case of O blood group, we have both i's and in AB, we have IA and IB. So it just gives you feel that in the blood groups, we have on one hand we are looking at what type of carbohydrates are present on the RBCs which is A, B, AB, or O.

And it is possible only one of the copy let you dominant. One of them is recessive, still it is going to show you the dominant property or in this case, both have to be recessive. So this is how in genetic terms, you can think about all type of blood groups. Now what is interesting that, you know, people can still derive the blood groups by doing certain reactions build on antigen and antibodies and interestingly we are going to perform that experiment in the class today shortly where, you know, you can challenge biology and concepts that it always was true or not.

If you already know your blood group and we will take your blood group and we will do those reactions and try to tell you what your blood groups are with a very simple logic of how an antibody reacts with an antigen and if that reaction happens, then you will see some sort of agglutination or some particles will be formed. So let us continue on this.

Let us kind of look at the situation where if the parents are, you know, continuing different alleles which are A, B and O type and just imagine the way we have done a normal crosses. So parent alleles are A, B, or O and then you do another, you know, so father and mother, both are having A, B and O alleles. Can you do that cross and then say that, you know, what are all possible genotype of those children for the blood groups possible, right. This is how you will see.

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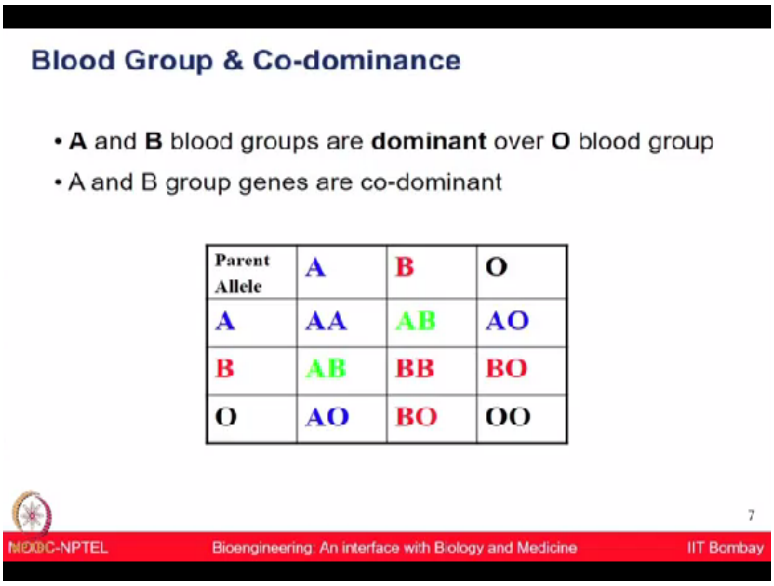

So you can now find out from this cross that what are the possible blood groups of these children. So now some of my TAs will show you how to determine human blood type based on simple experiment. Observe each step very carefully. This is based on very simple concepts of biology. If you have this kit at home, you can do this yourself. Although it is not advised to do that without experts but conceptually, you can understand and you can do this thing yourself.

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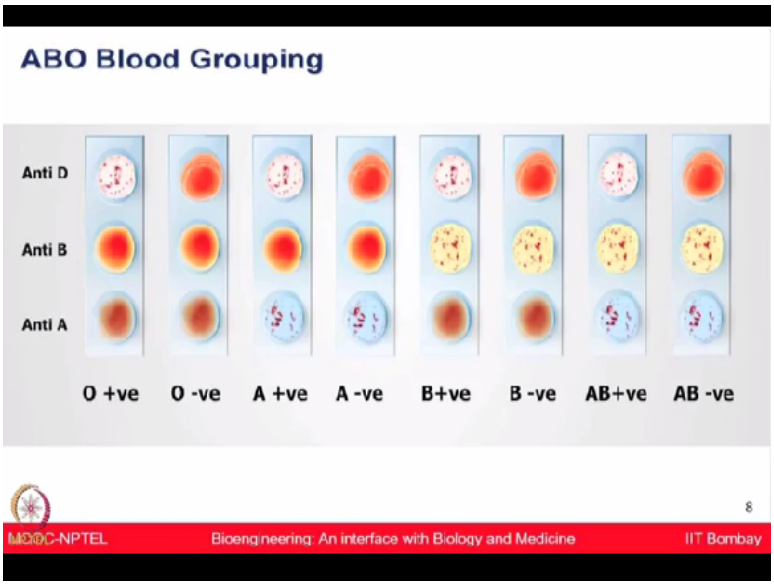

And it is very simple which shows elegantly how accurately biological experiments work if you are looking at antigen and antibody interaction and those reactions are always so accurate.

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Blood group typing is a type of experiment in which we determine the blood group of an individual. Karl Landsteiner discovered this ABO blood grouping system which is based on the fact that the surface of RBCs are covered with glycoproteins which are known as antigens. So according to this system, there are 4 types of blood groups. Blood group A, blood group B, blood group AB and blood group O.

Here we are going to conduct an experiment in which we are going to determine blood group of this blood sample. This is the pipette and I am going to pour 3 drops of blood on this slide. This is anti A. I am going to pour 1 drop of anti A to the first drop of blood. Anti B to the second drop

of blood. Anti D which is used to determine the Rh antigen on the blood surface. I am going to add anti D on the third drop of blood.

Now I am going to mix these. We have to make sure that while mixing we use a new toothpick each time. In this sample, anti B shows agglutination. This means the blood group is B. Anti B forms agglutination with the surface glycoproteins of this blood group. Also we can see agglutination in the third spot which denotes that this blood group is Rh positive. So as a whole this blood is B positive.

We have 3 antibodies. Let us say when we add antibody a, that antibody is going to react with antigen A. So in case of blood group A or in case of blood group AB, you are going to see this reaction, agglutination reaction to happen which is going to make these kind of particles. It will not happen for the O or the B. Now when you add antibody B, you will see the reactions happening either with the B or with the AB.

So so far we are not able to tell either positive or negative. It is just whether it is A or B, right. So that information you can get. Or if it is not reacting with both antibodies, then you can say it is O. If it is reacting with both of them, then you are saying it is AB. Then how you derive these 4, then by looking at the reactions of 2 antibodies. Now if you want to look at are their Rhesus factor positive or negative, so you are adding third antibody anti D.

And that antibody if it shows reaction positive, then you say it is O positive or O negative or A positive A negative, B positive B negative, AB positive AB negative. Now let us solve few problems based on this new concept that is deviation from Mendelian genetics. This section will mainly focus on problem sums which involves epistaxis and ABO blood grouping. My TA will solve some of these sums for you.

But please do try it out yourself. But before we proceed with problem sums, let us check if you have understood the concept clearly. Please mark the most appropriate answer based on what you have already learnt in the last few classes.

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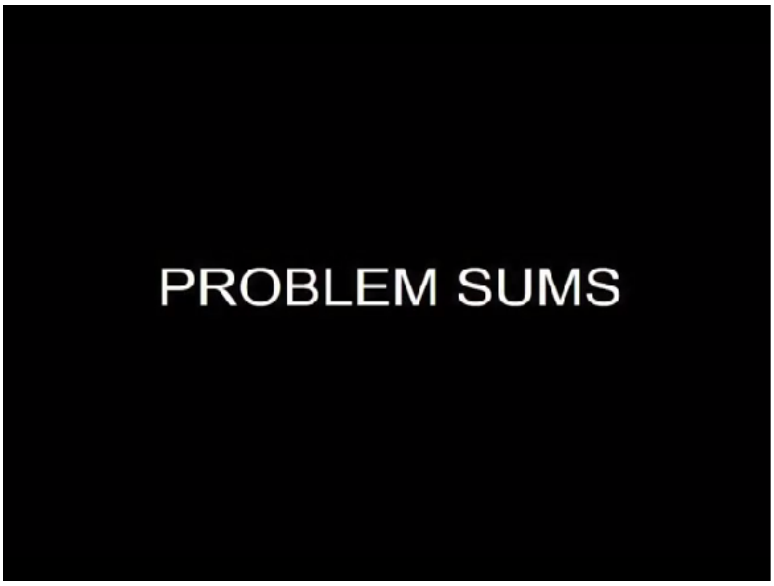

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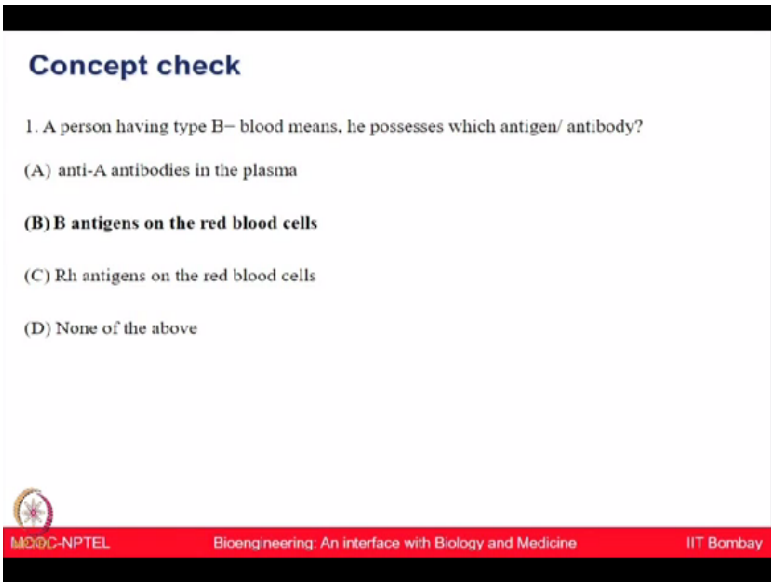

A person having type B- blood means, he possesses which antigen or antibody?

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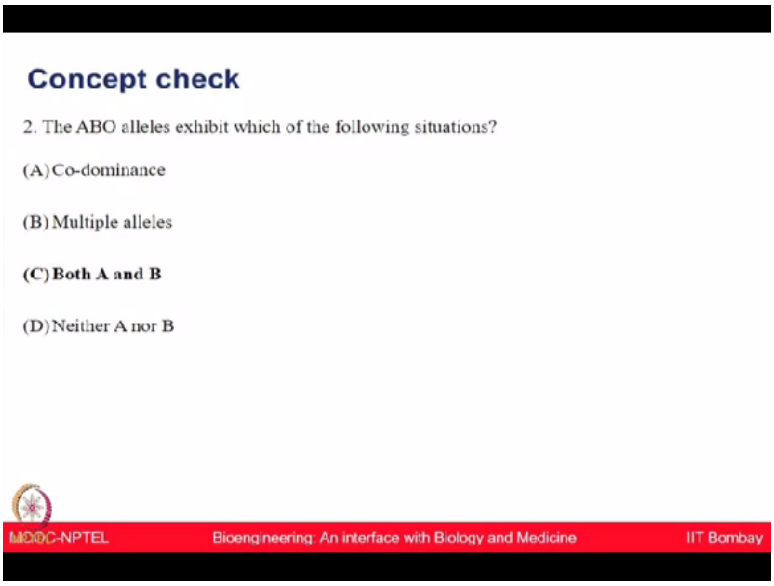

The ABO alleles exhibit which of the following situations?

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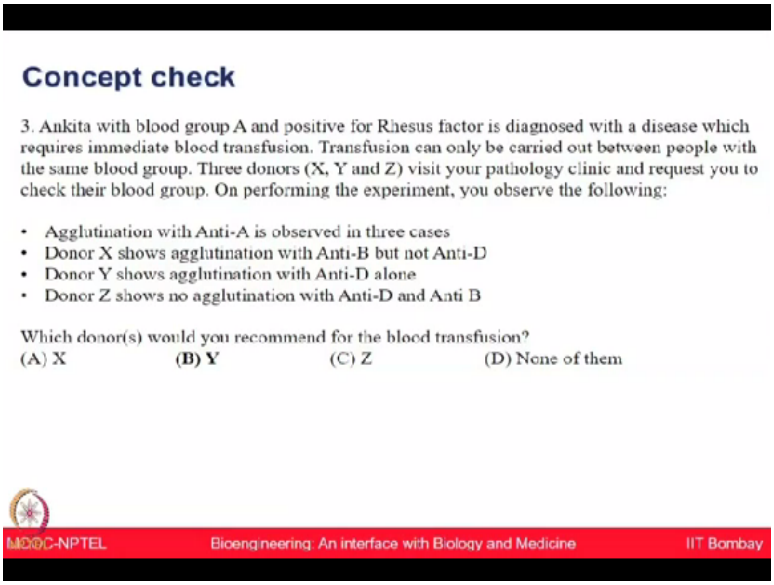

Ankita with blood group A and positive for Rhesus factor is diagnosed with a disease which requires immediate blood transfusion. Transfusion can only be carried out between people with the same blood group. Three donors X, Y and Z visit your pathology clinic and request you to check their blood group.

On performing the experiment, you observe the following. Agglutination with anti A is observed in 3 cases. Donor X shows agglutination with Anti B but not anti D. Donor Y shows agglutination with anti D alone. Donor Z shows no agglutination with anti D and Anti B. Which

donor would you recommend for the blood transfusion?

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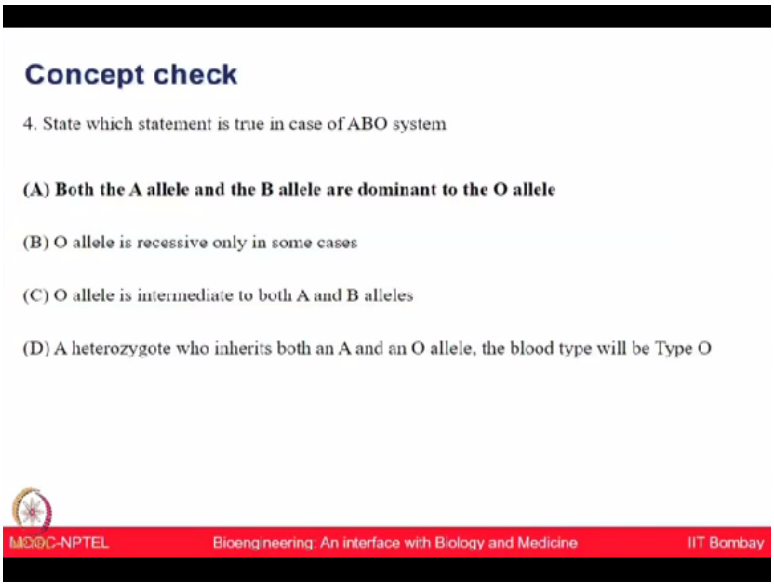

State which statement is true in case of ABO system.

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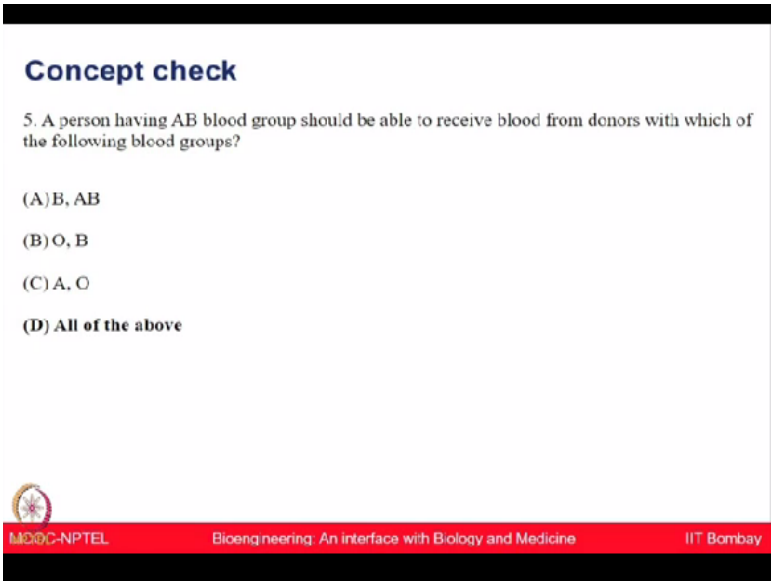

A person having AB blood group should be able to receive blood from donors with which of the following blood groups?

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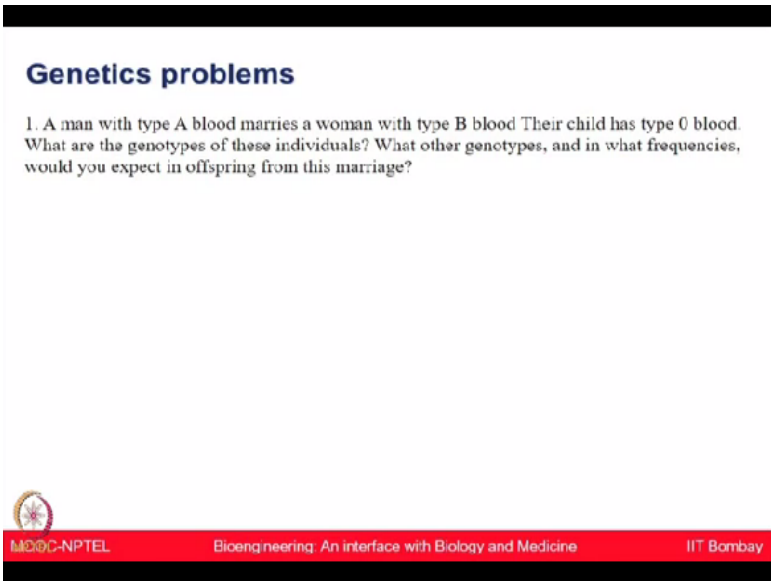

Let us now do some genetics problems. A man with type A blood marries a woman with type B blood. Their child has type O blood. What are the genotypes of these individuals? What other genotypes and in what frequencies, would you expect in the offspring from this marriage?

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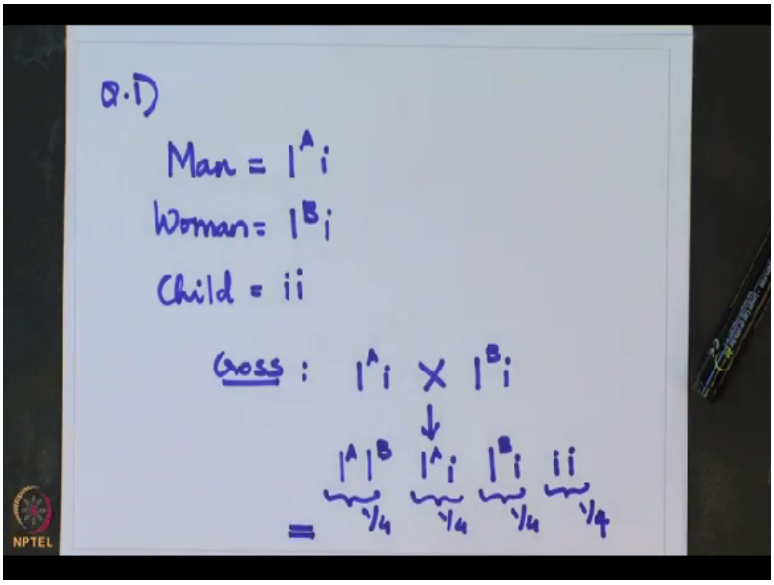

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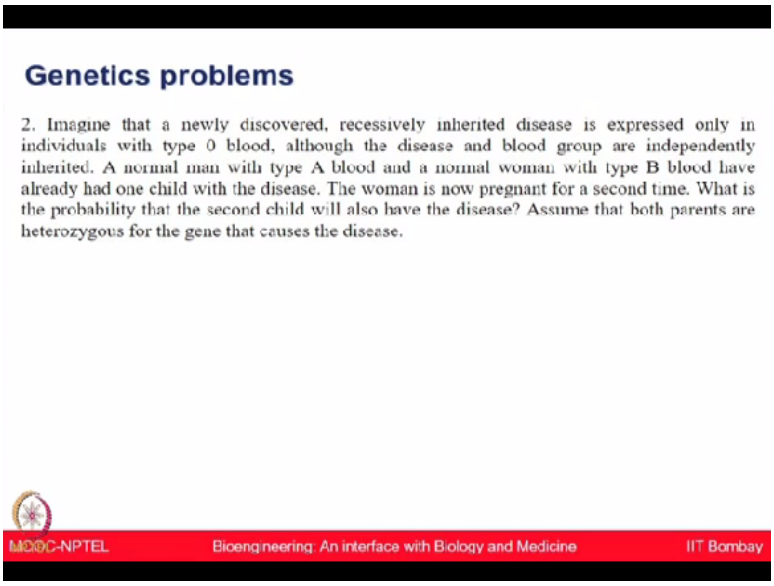

Let us now do second problem. Imagine that a newly discovered, recessively inherited disease is expressed only in individuals with type O blood, although the disease and blood group are independently inherited. A normal man with type A blood and a normal woman with type B blood have already had 1 child with the disease. The woman is now pregnant for a second time. What is the probability that the second child will also have the disease? Assume that both the parents are heterozygous for the gene which causes the disease.

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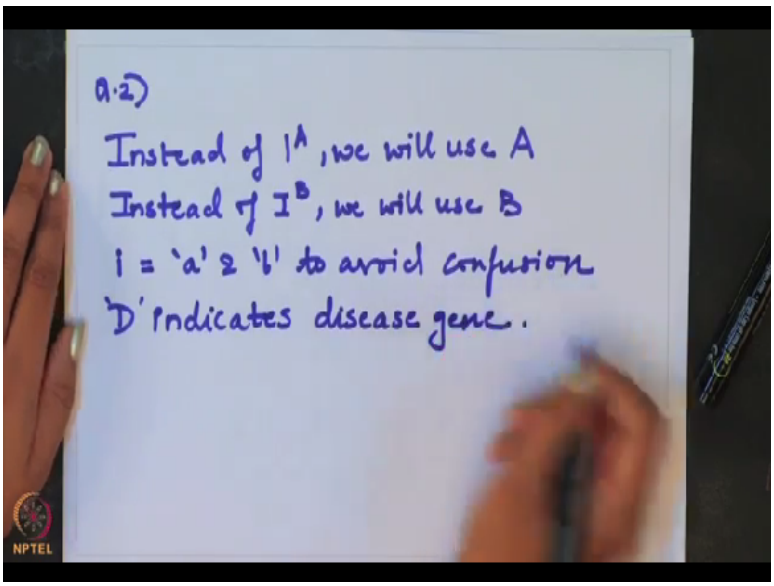

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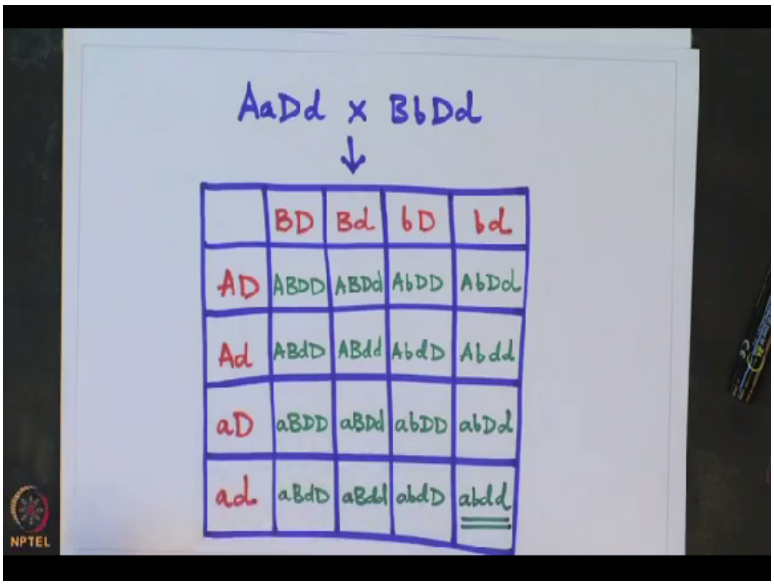

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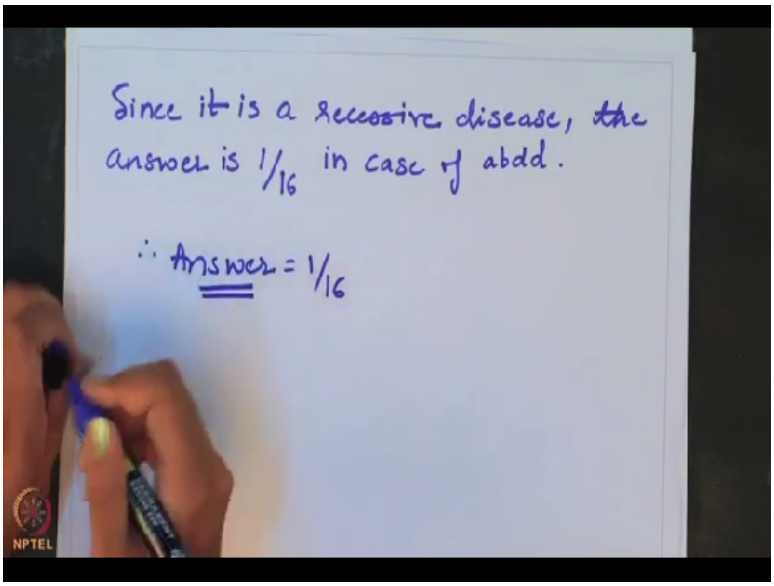

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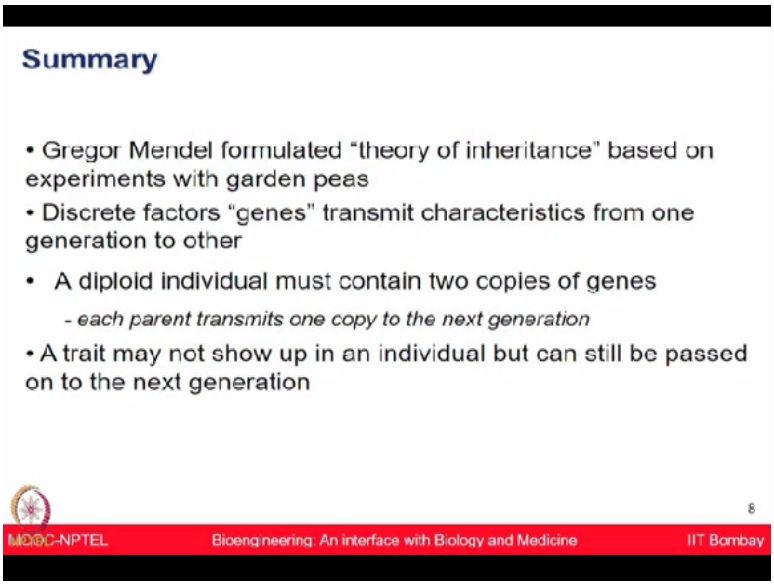

In summary, today we talked about Gregor Mendel and how he formulated the theory of inheritance based on experiments which he performed on garden peas. Mendel described genes as discrete factors and how these factors are transmitted, their characteristics from one generation to the next generation. A diploid individual must contain 2 copies of genes which it inherits from both the parents and each parent transmits 1 copy to the next generation.

A trait may not show up in an individual but it can still be passed on to the next generation. We will talk about genetics and rules of probability in next lecture. Thank you.

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