Biology for Engineers and other Non-Biologists
Professor G.K.Suraishkumar
Department of Biotechnology
Indian Institute of Technology, Madras
Lecture Number 17
Mendelian Genetics: Mendelian Inheritance Principles

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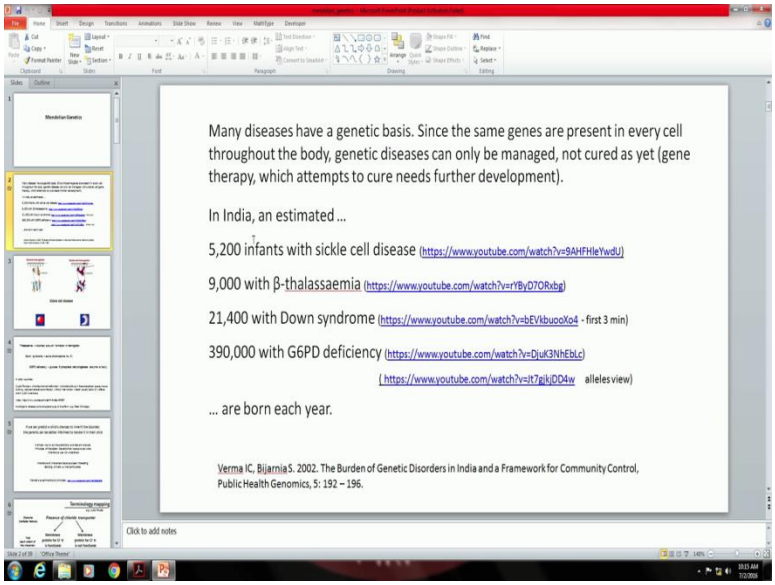

Welcome to the set of lectures on Mendelian Genetics. We saw that many diseases have a genetic basis; for example in our country, sickle cell anaemia or sickle cell disease, beta thalassaemia, Down syndrome, glucose 6 phosphate dehydrogenase deficiency are all occurring genetic diseases and in other countries such as the US, cystic fibrosis is a widely occurring genetic disorder. So our (Huntings) Huntington's disease and Achondroplasia and so on and so forth; these are just examples.

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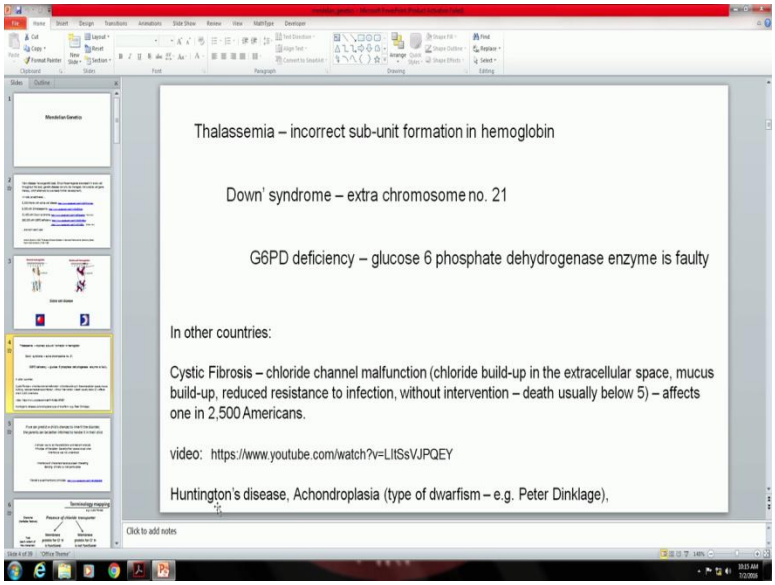

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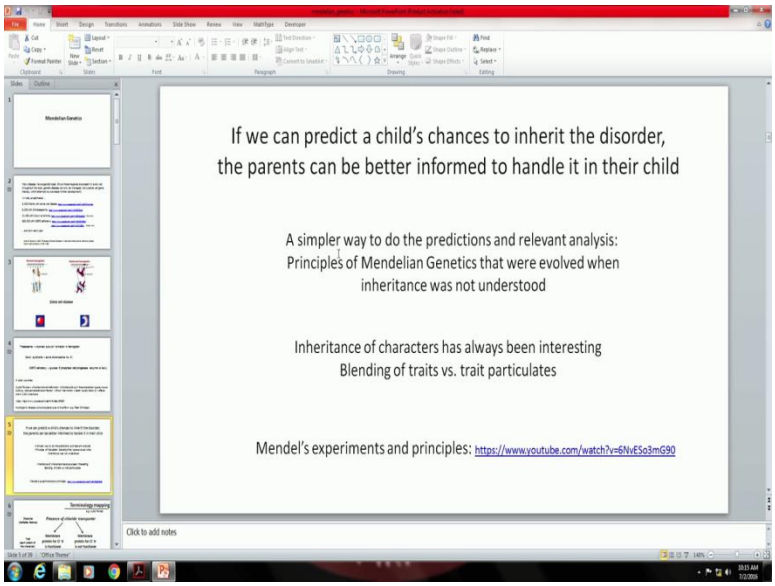

Our overall aim was that if we can predict a childs chances to inherit the disorder, the parents can be better informed to handle it in their child, we know quite a bit about the way the information is inherited or the (ca), the characteristics are inherited and so on, whereas for this kind of an aim, the principles of Mendelian Genetics which were developed about a century and a half ago, or more than a century ago, are very convenient to use; and that is the reason why we are looking at Mendelian Genetics here. I had pointed out I had pointed this video to you, please take a look

look at this video, if you have not already done so; slightly longish, but a very nice video on Mendel's experiments and principles.

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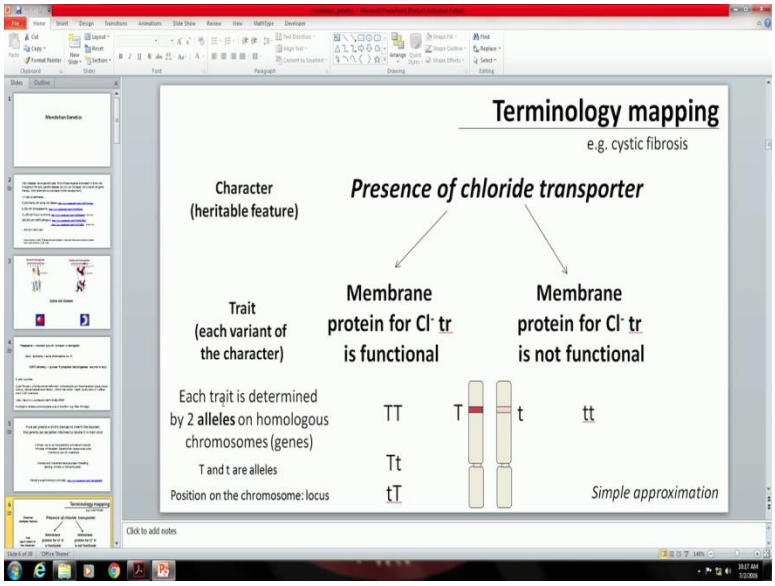

Before we went into Mendel's experiments, we needed to map whatever terminology that we use nowadays in the context of genetics to the equivalent terminology that goes with a Mendelian analysis of inheritance; for example, we had taken cystic fibrosis and we said presence of a chloride transporter, this is rather molecular in nature, it could be somebody being tall and short and so on so forth, that could also be and we said that this was a character which is an (inheritab) which was a heritable feature, and the presence of the chloride transporter, it could be of two kinds, if the membrane protein for chloride transporter is functional then it is fine; if the membrane transporter for chloride function, chloride transporter is not functional, then we get the disease.

Therefore there are two traits, the functionality and non functionality and these two traits are variance of this character and that is the terminology here. In other words, in terms of classic terminology; tall and short, these are the traits of the character height. Then we said that each trait is determined by two alleles on homogenous, homologous chromosomes which are the equivalent of genes; for example, this could be determined by TT capital that, capital T small t, small t capital T or small that, depending on what is present on these two loci, locus here, these two loci of the homologous chromosomes.

If both capital Ts are present, then the membrane protein for chloride transporter is functional; if at least one of them is capital T, then it is functional; if both are small, then it is non functional. This is the mapping between a molecular aspect that we know of nowadays to the olden times, the Mendelian times, when nothing of this was known, and it is very insightful to come up with this kind of a model to explain inheritance. And as you can see, this is a simple approximation, there are various different diseases that do not follow this approximation, we will first look at this approximation which is very useful and then I will extend that to show you the differences.

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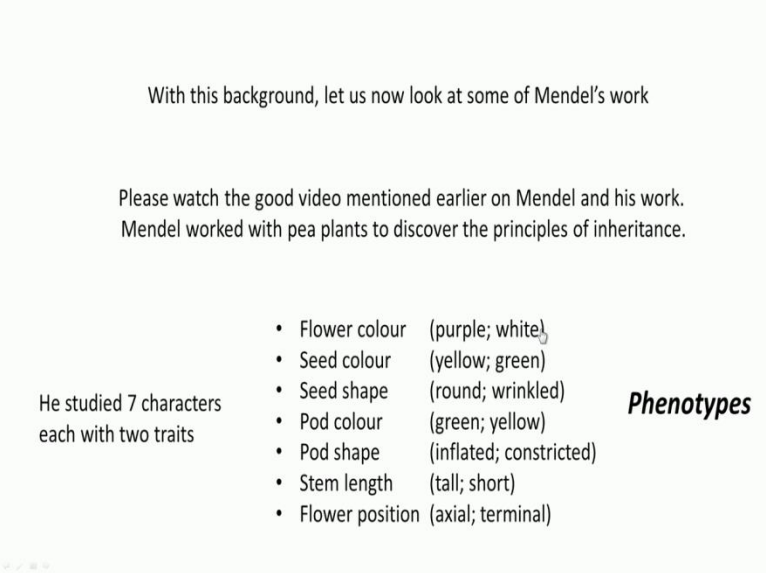

I think this is where we stopped last time, so let us go further with the lecture material of this lecture. We are going to review, we are going to see some of Mendel's work. Again let me emphasize the watching of this video. Mendel worked with pea plants to discover the principles of inheritance, you know, the very first lecture, the introductory lecture, I had mentioned about studying something for the sake of it, studying something because we want to understand something that exists around us. This study was one such kind and look at where it has led to.

Coming back to Mendel's work, Mendel studied seven characters, each of which had two traits. The characters that he studied were flower colour, seed colour, seed shape, pod colour, pod shape, stem length and the flower position. Let us go further to see, to explain this and these characters, flower colour, seed colour, seed shape, pod colour, pod shape, stem length and flower

positions are called ‘phenotypes’; for example, the flower colour could be either purple or white; both are phenotypes, flower colour is a phenotype and so on.

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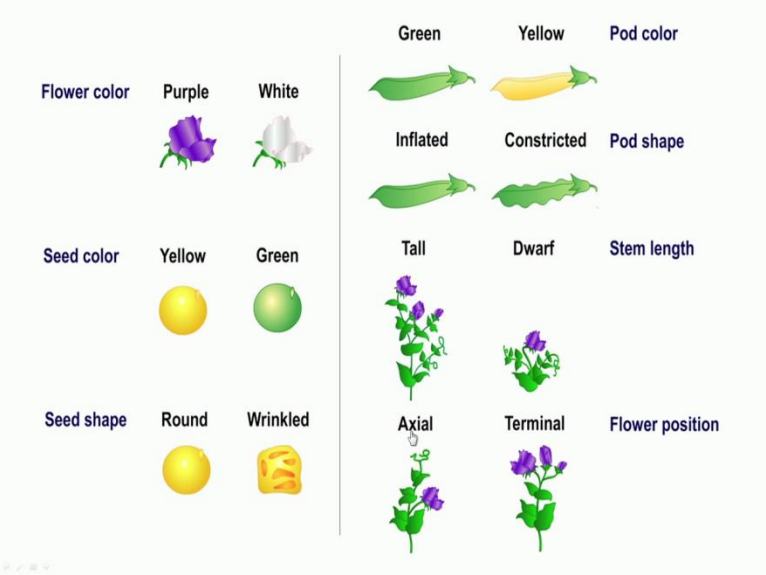

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Here, we see it in a pictorial form in the particular case of pea plants; the flower colour would either be purple or white; the seed colour would either be yellow or green; the seed shape would be round or wrinkled, by round it is actually smooth, round or wrinkled; the pod colour could either be green or yellow; the pod shape could be either inflated or constricted; the stem length

could be either tall or dwarf; and the flower position could be either axial or at the end, terminal, okay?

So these were the characters that Mendel studied, most of these characters were rather simple, it just happened by chance that they were simple and therefore the basic principles of inheritance could be became amenable to study by Mendel; he could come up with the basic clause of inheritance, most of which is, was applicable for simple inheritance. Now let us look at one character, okay, which is flower colour. Mendel started hi experiments with true (bleedi) breeding plants. What does true (bleedi) breeding mean?

It means, in the context of flower colour, the same (colo) of flower results in all the offspring in all the generations, okay; which means it is true breeding amongst itself and therefore if it is purple; if the flower colour is purple it is purple throughout in all the generations forward; if it is white, it is white in all the generations forward. Such plants are called true breeding plants and Mendel was careful to spend the time to achieve true, to make sure that they were indeed true breeding plants before he started out his experiments.

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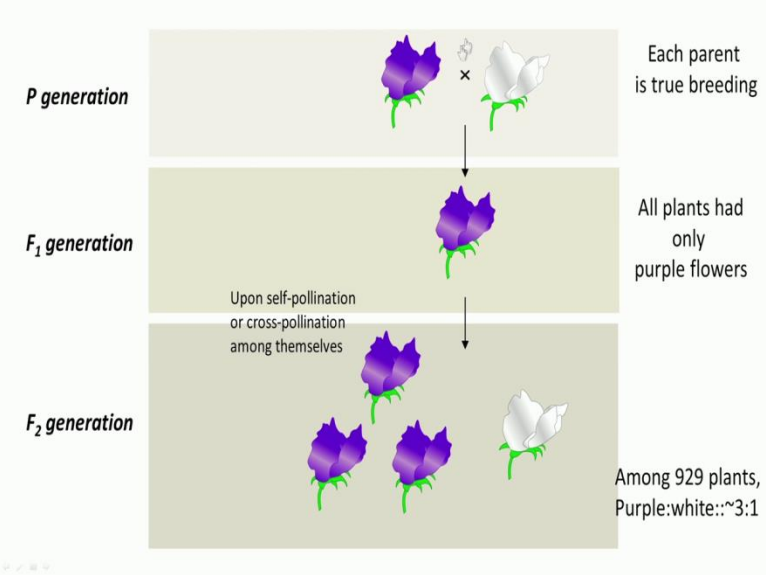

So when he started out with true breeding plants, the ones that yielded purple colour flowers and the ones that yielded white (cover), white colour flowers, and he crossed them with each other. The pollen from and the female part of the plant were made to interact in different ways and this cross resulted in a purple flower in all the plants of the first generation; this is the parent

generation; the first generation had only (puple) purple flowers, there was no sign of the white flower at all, and there was nothing in between; it was either purple, rather in this case, it was purple or white, and in the F1 generation, it was all purple. This was the observation, and he had observed this over a large number of plants and he did a statistical analysis to find that a statistical analysis of course does not mean much here; he did statistical analysis over the entire set of experiments to come up with these various findings.

Here it does not make a difference; here everything had only (pa) purple flowers. And then what he did was he either self pollinated or (cross) cross pollinated the purple flowers amongst themselves, okay, this cross with another purple flower of the F1 generation itself. So when that happened, in the F2 generation, something strange happened. Three fourths of the flowers, he had studied about nine hundred and something plants, yeah here, among nine twenty nine plants, about three fourths were purple, and one fourth was white approximately, in a very large number of plants. Okay. So the white colour which was missed in the first generation made an appearance in the second generation; that is what he found.

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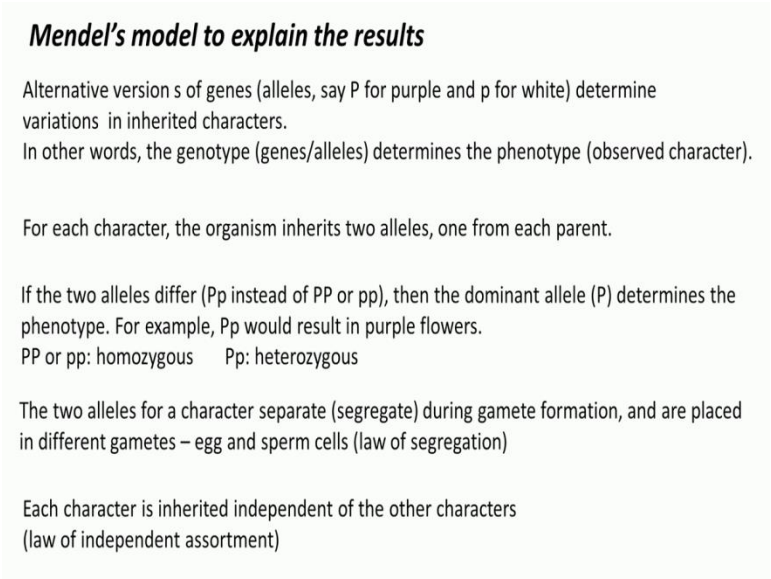

So after a lot of experiments with a lot of single characteristics, dual characteristics and so on so forth, Mendel came up with a model to explain the results which can be given as follows: alternative versions of genes or alleles, say capital P for purple and small p for white determine the variations in inherited characters, this is the first aspect of the model; in other words the

genotype, (gele), genes or alleles determine the phenotype which is the observed character. The P determines the purple and the small p determines whether it is white.

For each character, the organism inherits two alleles, one from each parent. If the two alleles differ, you know, capital P small p, instead of both being capital P or both being small p, the dominant allele P determines the phenotype, okay? For example, if there is a capital P and a small p, the trait of capital P is what would be seen. In other words, this would result in purple flowers. In terms of terminology, if both are the same, and either both capitals or both smalls, then they are considered homo, same, zygous. okay, were the same; and whereas if they are different, they are called hetero, different, zygous, okay?

Homozygous and heterozygous, these are commonly used terms, it is nice to know. If both are the same, homo, so homozygous, if both are different, hetero, so heterozygous. The two alleles for a character separate out or segregate during gamete formation, right, you would have gone through meiosis, you would have studied meiosis, so you know what a gamete is; this is what results during, in the cell division, that is gone through by the germ cells. Here we are talking about the separation of the two alleles, and what Mendel said was that they segregate during gamete formation and are placed in different gametes, the egg and the sperm cells. Okay? And this is called the law of segregation.

The two alleles separate, they are placed in separate gametes; and each character, for example flower colour, seed colour, seed shape, these are all different characters. Each character is inherited independent of the other characters. Right now we are looking at only one character, but all characters are being inherited simultaneously. That we all know. And what Mendel said by his studies was that each character is inherited independent of the other characters, and this is called the law of independent assortment. These two are supposed to be major contributions; the ‘law of segregation’ and the ‘law of independent assortment’. Why this is so will become clear very soon. It blends itself to very easy analysis.

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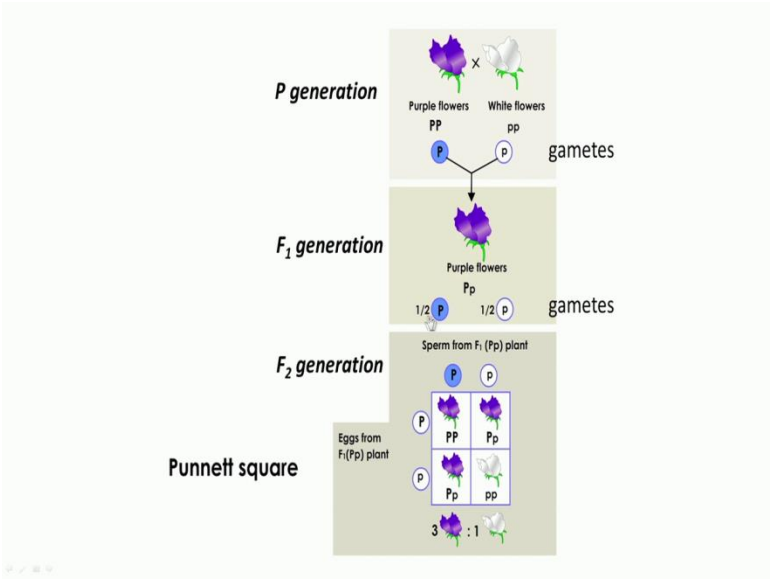

Therefore, if you look at the earlier experiment again, the P generation had purple flowers, white flowers. The purple flower, these were true breeding the true breeding varieties and therefore the purple flowers means both are capital P, and the white flowers means both are small p, okay? In other words, there is no heterozygosity here. So so the gametes would have both capital P, in this case, and both small ps in this case; and upon fertilization, the genotype is capital P small p, which are, which results in a purple flower because capital P is dominant, right?

And the gametes from this would have, half of them would have capital P, and half of them would have small p, and therefore in the F2 generation, if you look at all possibilities, this is let us say, from the sperm of the plant, and this is from the eggs of the plant; capital P small p, capital P small p, and the combinations would be both capital Ps, the here it will be capital P small p, here it will be capital P yeah, this is the eggs, which is , this is Pp, both are P; capital P small p capital P small p, and therefore this will be capital P small p, and this will be both small ps, okay?

If atleast one is a capital P, then it will result in a purple flower because it is dominant, and therefore you have three out of four being purple and one out of four being white, okay? This kind of an analysis where you look at all possibilities by using this kind of a square with the gametes from the sperm and the gamete from the eggs is called a 'Punnett Square', okay? This is heavily used in this kind of analysis. It's rather nice if you are not dealing with very complex

situations; to characters it is quite good to do, it gives you a visual feel for what is happening, okay? Whereas, if you have, if you are comfortable with probabilities, that is a much easier way to do, okay? We will, we will look at that in a little while.

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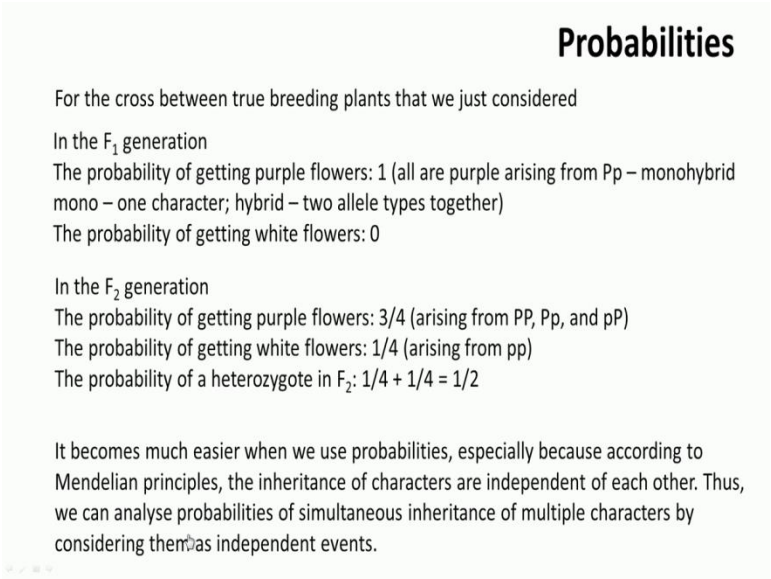

Now let us look at probabilities. It is, as I said, if you look at it in terms of (probabil) probabilities, if you are comfortable that way, then it becomes much easier to do the analysis. Let us apply this or let us look at whatever we did just now, the cross that we did just now, in terms of probabilities. For the cross between two breeding plants that we just considered in the F1generation, the probability of getting purple flowers was one; all were purple, either resulting from , rather as a rising from capital P small p, okay? So monohybrid, mono is one character, hybrid means it is a mixture of two allele types. And the probability of getting white flowers is zero.

Whereas in the F2 generation, the probability of getting purple flowers is three-fourth arising from either homozygous condition or heterozygous condition; capital P capital P homozygous, capital P small p in both the, both these kinds is heterozygous. And the probability of getting white flowers was one-fourth, arising from only small p small p of all the possible combinations. And therefore, the probability of getting a heterozygote in F2, right, is one-fourth plus one-fourth, which is half and so on. You could do various different calculations of this kind in terms of probabilities; you can find the probability of various different things.

So you don't have to go and draw squares which is somewhat elaborate and so on, if one is comfortable with using probabilities. It becomes much easier when we use probabilities, especially because, according to Mendelian principles, the inheritance of characters are independent of each other, okay, law of independence. Thus, we can analyze probabilities of simultaneous inheritance of multiple characters by considering them as independent events, okay? We become clearer here when we consider two characters.

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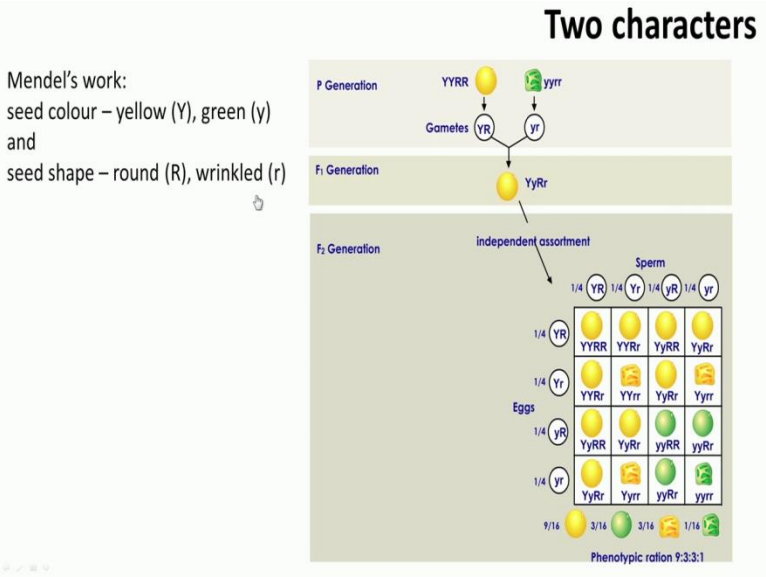

Now let us see the way the Mendel, the way Mendel saw it. For that, let us look at seed colour, yellow or green, yellow capital Y, green small y and seed shape round capital R, wrinkled small r being inherited together, or being (con) considered together, okay? Same kind of a cross, true breeding, therefore capital Y capital Y, capital Y capital Y, capital R capital R, two characters here, seed colour and seed shape, and small y small y, small r small r. The gametes in this case would be capital Y capital R, the gametes in this case would be small y small r; upon fertilization it is going to be completely heterozygous, a (he) yeah, a capital Y small y capital R small r and then they are going to be in a, independently inherited or independently assorted.

Therefore the seed colour would be independent of seed shape, the inheritance of seed colour would be independent of the inheritance of the seed shape, that is what it means. If we drop Punnett Square here of all the possibilities, you can see if you count, that nine out of the sixteen possibilities would be yellow and round, both dominant characters; three out of sixteen would be

round and green; three out of sixteen would be yellow and wrinkled and one out of sixteen would be green and wrinkled. This is how it happens with two characters which we can get an idea by using the Punnett Squares here.

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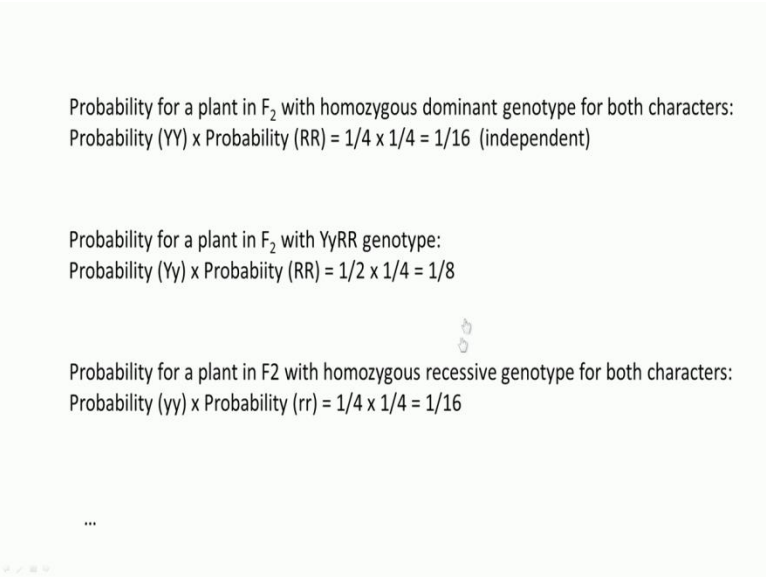

In terms of probabilities; probability for a plant in F2 with homozygous dominant genotype for both characters. What does it mean? Homozygous dominant genotype. 'Homozygous' means both should be the same; 'dominant' means, in our terminology, both should be capital, right? So in other words, probability of capital Y capital Y, and small capital R capital R. We know that the (probabili) probability of capital Y capital Y is one-fourth, and the probability of capital R capital R is one-fourth, and therefore, since these are (inhe) independently inherited, independent assortment, you can multiply these, one-fourth one-fourth, that is one-sixteenth, okay. Independent events, you can multiply the probabilities.

Now the (pro) probability of a plant in F2 with capital Y small y, capital R capital R genotype, okay; these two are independent; therefore the probability of capital Y small y into the probability of capital R capital R, half into one-fourth that is one-eighth. The (pro) one more thing, just to illustrate how you could use this, it becomes very easy if you are comfortable with probabilities. The probability of a plant in F2 with homozygous recessive genotype for both characters, 'homozygous' both the same, 'recessive' both small letters in our terminology. In other words, probability of small y small y, small r small r. We know that this is one-fourth this

is one-fourth, and therefore, the probability in F2 with, or for a plant in F2 with homozygous recessive genotype would be one-fourth into one-fourth, that is one-sixteenth, and so on and so forth. You could do a lot of possibility probability calculations with this approach. You could also bring in three characters and so on and so forth; four characters and so on, then you can work this out.

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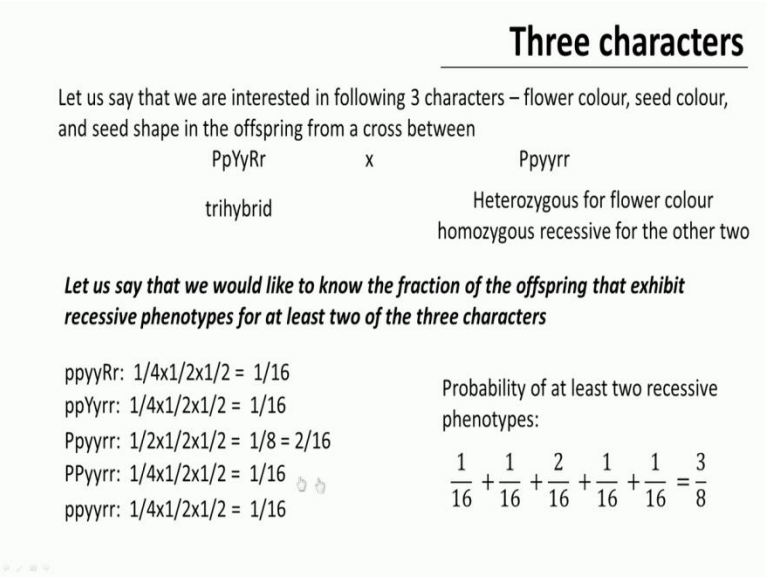

There is three characters here. Let us say that we are interested in the following three characters: flower colour, seed colour and seed shape in the offspring from a cross between capital P small p, capital Y small y, capital R small r, and this is heterozygous in P, whereas homozygous in Y and R. So this is a trihyrid, and this is heterozygous for the flower colour, whereas homozygous is recessive for the other two.

And let us say that we would like to know the fraction of the offspring that exhibit recessive phenotypes for atleast two of the three characters. This is what we are interested in. Okay. If you go through the Punnett Square, it is some amount of work, whereas if you know probability, you can (qui) quickly write down this, okay? Two should be, two of the characters should be recessive, okay? Therefore it could be either p-recessive, y-recessive or r-recessive. , I am sorry; exhibit recessive phenotypes for atleast two, okay? So taking two at a time. So in this case P and Y are recessive, the probability is one-fourth for P being recessive, half for Y being recessive,

you can go back and check that, and that has to be combined with this, if only two are going to be recessive, this has to be dominant.

Therefore capital R small r is one possibility, and this is half. Multiply all these things together because of the law of independent assortment, you get one-sixteenth. This is one of the possibilities in which this offspring that exhibits recessive phenotypes for atleast two of the three characters would arise. The other possibility is P and R being recessive. So if you work out the probabilities for this combination, one-fourth, half and half, multiply all together, one-sixteenth. Another possibility is Y and R being recessive, that is and the first one being heterozygous; so half, half and half, that turns out to be one-eighth, just to keep the denominator the same, let us write it as two-sixteenth.

Here is the other possibility, Y and R being recessive, both homozygous, whereas this one is dominant, (bo) homozygous dominant; this is heterozygous dominant, this is homozygous dominant, and one-fourth half half multiply all, one-sixteenth, and this is all recessive; it says atleast, for atleast two or three characters, two recessive phenotypes, so all three can be recessive. So that (proba), those probabilities are one-fourth, half and half, for small p small p, small y small why, and small r small r, multiply all together, one-sixteenth. Right? So, in all these ways, you could get offspring that exhibit recessive phenotypes for atleast two of the three characters.

So we just have to add all these things together, total probability; so probability of atleast two recessive phenotypes, it is just addition of all these probabilities that turns out to be six by sixteen or three by eight. So we, a lot of probabilities this way are found and they turn out to be useful as we will see in some ways later on when we discuss diseases. If you look at what has happened, in terms of what is happening during meiosis, this is what happens.

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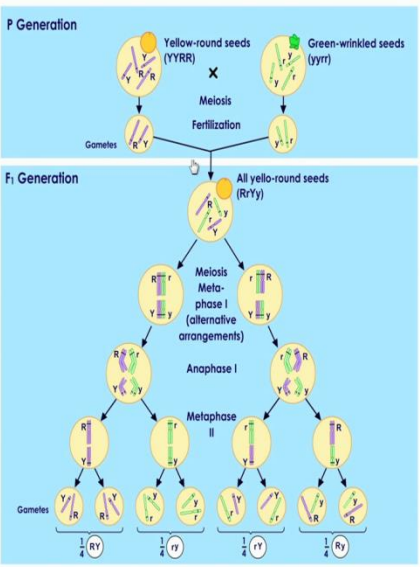

The P-generation here, I think slightly different example is considered; yellow round seeds, the these are two traits being considered together, that is fine and these are green wrinkled seeds, yellow round seeds true breeding, therefore capital Y capital Y capital Y small, capital, capital Y capital R capital R, and green and wrinkled would be small y small y small r small r.

During meiosis they form gametes. These gametes would only be capital Y capital R; these gametes would only small y small r and upon fertilization, these are the two possibilities, let us look at them one by one. they get mixed up and by the way they arrange themselves, it would be capital R small r here, capital Y small y here, please go and take a look at what happens during meiosis, especially during metaphase one and anaphase. In the anaphase these start separating out and metaphase two, you have these gametes being formed, capital R capital Y and small r small y and therefore the gametes here would all result in either capital Y capital R, these two, or small y small r, okay?

Alternatively, if the arrangement is like this, small r capital Y capital R small y, then during anaphase, they segregate out resulting in small r capital Y, capital R small y and small r capital Y, small r capital Y here, capital R small y capital R small y here. Therefore, one-fourth, you know two out of eight is capital R capital Y, one-fourth is small r small y, one-fourth is small r capital Y, and one-fourth is capital R small y. This is essentially what has resulted from this kind of a segregation. This will become clearer when we look at two things together, but this gives

you the molecular basis for the Punnett Square itself, in this case two characters, but you could also look at it in terms of one character. I think this is right time to close this lecture.

Here we saw Mendel's experiments with pea plants, their results and analysis of those results using simple probabilities and the law of independent assortment that Mendel found. When we move forward, we will take things further towards diseases and see the use of 'Mendelian genetics' to be to predict diseases. See you in the next lecture.