#### Bioengineering: An Interface with Biology and Medicine Prof. Sanjeeva Srivastava Department of Biosciences and Bioengineering Indian Institute of Technology - Bombay

### Lecture – 17 Genetics-II

Welcome to MOOC-NPTEL course on bioengineering, an interface with biology and medicine. Today, we will discuss some examples of Mendelian genetics. You will see how many human traits follow Mendelian patterns of inheritance but how these patterns are often more complex than just predicted by simple Mendelian genetics.

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So there are many examples of Mendelian genetics which we can study and I must say that, you know, the Mendelian rules, Mendelian laws were made based on the pea plants with very limited conditions. So we had 1 gene 2 alleles and we are, or we are talking about 2 genes and 4 alleles kind of properties.

But many times when we talk about human characteristics, things are much more complicated, much more complex. And at that time, the characteristics are governed from many genes, polygenic in nature. So it may not be always true that, you know, every inheritance pattern is going to follow Mendelian's law. It may not be always true.

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But there are many human traits which will still follow Mendelian rule and people have done the pedigree analysis.

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	Pedigree analysis	
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It means in the same family if you look at, you know, your grandfathers and the grandsons, their entire, you know, properties, are their certain characteristics which are appearing and disappearing and reappearing kind of stuff.

(Refer Slide Time: 01:30)



So that is a pedigree analysis when you are looking at a family history for particular trait and then you are assembling that information and trying to derive some sort of family tree, a pedigree information from those traits. So many of the human traits for example, one is known as widow's peak which is something shown here.

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So this property is actually, you know, the sharp hair line is known as widow's peak which is observed in many families and these are dominant traits which are actually being followed in those women and they are always following the Mendelian ways of inheritance, Mendelian laws of inheritance.

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• Attached earlobe	o follow Mendelian genetics (recessive trait)	
Key ☐ Male ☐ Affected male → Fernale Affected female	Mating Offspring, in birth order (first-born on left)	
	Ist generation (grandparents) 2nd generation (parents, aurts, and uncles) FF  or  E'  ff  ff	
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There are certain characteristics like if you look at the earlobe in this case here. So this attached earlobe or then the free earlobe, you will see some variations like this and then these are some of the recessive characteristics which are also found in some family and they also obey Mendelian laws of inheritance.

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So but as I mentioned the inheritance patterns are sometimes very complex. They are not as simple as just looking at a pea plant and 1 gene at a time. They are governed from many genes and many complex information. Those are polygenic in nature. So Mendel in some way was actually lucky because he was only looking at 1 thing at a time.

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#### **Complex Inheritance Patterns**

• In Mendelian genetics each character was determined by one gene, for which there are only two alleles (completely dominant or completely recessive)

• Relationship between genotype and phenotype are not always as simple!



And then he derived his conclusions based on those relationship between genotype and phenotype. Ideally is not as simple as just looking at their 1 genetic content.

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Degrees of Dominance	
<ul> <li><u>Complete dominance</u> occurs when phenotypes of th heterozygote and dominant homozygote are identica</li> </ul>	e I
<ul> <li>Incomplete dominance the phenotype of F<sub>1</sub> hybrids is somewhere between the phenotypes of the two pare varieties</li> </ul>	s ntal
<ul> <li><u>Co-dominance</u> two dominant alleles affect the pheno separately, distinguishable ways</li> </ul>	type
<u>(*)</u>	8
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So I will show you couple of examples where the dominant and recessive things may not be exactly following the same trends. So when we have, you know, the Mendelian kind of inheritance of complete dominance and recessive, that is known as complete dominant characteristics.

Sometime we have incomplete dominance, the phenotype of F1 hybrids. They are somewhere in between the phenotypes of the 2 parental phenotypes. Or the co-dominance, it means there are 2

dominant alleles. They are resulting into a third property which is not exactly same like any parents. So those are co-dominant allele or co-dominant properties, are some of the dominant alleles.

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They are for a particular characteristics always more common than the recessive alleles. **(Refer Slide Time: 04:01)** 



People have also looked at some of the frequency of dominant alleles. In this case look at the hand. So this is one of the dominant characteristic, especially lot of child born in US, they are having polydactyly, you know, multiple fingers, more than 5. So this extra finger or toes that is one of the characteristics which is example of polydactyly.

Recessive alleles in this case is the normal appearance with the 5 digits and is far more prevalent than the dominant allele. So is this because of natural selection. I think some of these, we will discuss in the context of evolution but just going to highlight that, you know, there are many variations in the human population available where one could start thinking about, you know, genetic principles.

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And some of these things one could think about at the single gene level and some of those are derived at the multiple gene level.

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PROBLEM SUMS BASED ON MENDELIAN GENETICS



We will now solve some problems based on the Mendelian genetics. Please try to solve them yourself first. Try to draw Punnett squares where where applicable. It will be easier to solve questions in that way. Also remember 2 Mendelian laws, law of segregation and law of independent assortment and the concept of inheritance.

The law of segregation suggest that 2 alleles for heritable trait separate from each other during gamete formation and form different gametes. The law of independent assortment states that 2 or more genes assorted independently and each pair of alleles segregate independently during the gamete formation. Please revise the basic terminologies like what is dominant, recessive, genotype, phenotype, alleles and traits.

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Let us solve few problems now. You should try yourself but a TA will also guide you to show the solutions. So let us start it now.

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Concept check	
<ol> <li>When true breeding plants self-pollinate, all the offsprings are of type(s)</li> </ol>	
(A)Different	
(B)Same	
(C)Two	
(D)Difficult to predict	
A	
(*)	11
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When true breeding plants self-pollinate, all the offsprings are of which type?

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Which of the following represents a test cross?

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Concept	check	
3. Two alleles for more common tra	leaf color are green (G) and orange (g). Green is dominant. O it. What is the phenotype of a homozygous dominant individu	range is the tal?
(A)Orange		
(B) Green		
(C) A combinatio	n of A and B	
(D) Yellow		
A common trait is dominant but	need not always be dominant and vice versa. For example, v NOT a common trait	Widow's peak
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Two alleles for leaf colour are green G and orange g. Green is dominant. Orange is the more common trait. What is the phenotype of the homozygous dominant individual?

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Genetics problems	
Q1. Self pollination of two pea plants heterozygous for flower position and sten length (AaTt) yields many seeds, of which 200 are planted. How many offspring would be predicted to be dwarf with terminal flowers?	n gs
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Self-pollination of 2 pea plants heterozygous for flower position and stem length AaTt yields many seeds, of which 200 are planted. How many offsprings would be predicted to be dwarf with terminal flowers?

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9.1 Aatt × Aatt Heterozygrus The satio will be Dway with terminal flowers 1.1 The actual result is not always exactly similar to what we get

Say suggestively you should not make the Punnett square in case of heterozygous crosses. Simply use 9:3:3:1 ratio. So according to the law of independent assortment, 1 out of 16 of the offsprings are predicted to be aatt or recessive for both characters. The actual result is likely to differ slightly from this value.

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Genetics	problems	
Q.2 A pea plant l homozygous for genotypic and ph	heterozygous for axial flowers $(Aa)$ is crossed with a plar terminal flowers $(aa)$ . Draw a Punnett square for this cro tenotypic ratios.	nt oss to predict
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A pea plant heterozygous for axial flowers Aa is crossed with a plant homozygous for terminal flowers aa. Draw a Punnett square for this cross to predict genotypic and phenotypic ratios.

(Refer Slide Time: 09:19)



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#### **Genetics** problems

Q.3 In some plants, a true-breeding, red-flowered strain gives all pink flowers when crossed with a white-flowered strain: RR (red) X rr (white) gives Rr (pink), lf seed shape is inherited (round or wrinkled) as it is in peas, what will be the ratios of genotypes and phenotypes of the F1 generation resulting from the following cross: round-red (true-breeding) X wrinkled-white? What will be the ratios in the F2 generation?

In this particular sum, one character belongs to a flower (flower colour) and the other belongs to a seed (seed shape).



In few plants, a true-breeding red-flowered strain gives all pink flowers when crossed with a white-flowered strain RR which is red crossed with rr which is white, gives Rr which is pink. If seed shape is inherited round or wrinkled as it is in peas, what will be the ratios of genotypes and phenotypes of the F1 generation resulting from the following cross which is round and red, true-breeding, crossed with wrinkled and white. What will be the ratios in the F2 generation?

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Q.3 RRX22 RR - F1 Ina Pink wss Red/White = R/2. Round/ Warinkled = 50 - For seed

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1	Now wossing FI,
12	SRSA × SRSA
	F27
	SR SZ SR SZ
	SR SSRR SSRL SSRL SSRL
	52 SSRA SSUL SOR SOR
18	S.R. SERR SEL SSRR SER
( <b>0</b> )	52 Sole Sou sole sore
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The phenotypic sation is : -6 around seeds with pink flowers 3 around seed with red flowers 3 sound, white 2 buindled, pink 1 wainkled, white 1 whinkled, sed . 6:3:3:2:1:1

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A man has 6 fingers on each hand. His wife and their daughters are normal. Remember that extra digits is a dominant trait. What fraction of this couple's children would be expected to have extra digits?

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Trait represented by D' D for Digits. Situation 2 Situation 1x Male × Female Male × Female Dd J DD dd dd xDd x dd Daughter

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Inspired from Mendel's experiment, now you want to perform some genetics experiments in your farm. If you have a plant called Vicia ervilia or Bitter vetch growing in your farm. This plant is usually short and bushy but the one growing in your farm is rather long. Now, assume that height in these plants is controlled by a single gene with short plants S being dominant to the long ones. So what is the genotype of your plant?

Could one of the parents of your plant have had a short phenotype? And if so, what would have been the genotype of that parent?

(Refer Slide Time: 22:50)



Let us say your friend help you to cross your plant with that of your neighbour's same Vicia plant

which are short and bushy. You found that half of the resulting offsprings are short while others are long. What could be the genotype of your neighbour's plant?

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Situation 1 55

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So in conclusions, now you are familiar with some of the examples of Mendelian genetics.

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In the next lecture, we will discuss some deviations from Mendelian genetics where you will understand that not everything in the world follows Mendelian rules. Thank you and see you next week.

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