Hello students. I am Puja Sakhalkar, Assistant Professor at Carmel College of Arts, Science and Commerce for Women, Nuvem. Today we are looking at the module 'Pleiotropy, penetrance and expressivity'. This is from Unit 2 of BOC 108 that is Mendelian genetics and its extension.

To give you an outline of this module, we will look at the concepts of pleiotropy, penetrance, expressivity and the effect of environment on penetrance and expressivity. This module explains the concept of pleiotropy, penetrance and expressivity using suitable examples. It makes you understand the effect of environment on penetrance and expressivity.

Now, let us look at the first concept that is pleiotropy. What is pleiotropy? Pleiotropy refers to the influence of a single gene on more than one phenotypic trait in an individual. So, what it means is that, a single gene can affect more than one character. This gene is called as the pleiotropic gene. Pleiotropic gene is a gene that is responsible for pleiotropism and the effects that are seen as a result of this are called as pleiotropic effects. Pleiotropy was first discovered by Gregor Mendel. He observed that in garden pea a single gene controls flower colour, seed colour coat and red spots on leaf axils. Some more examples of pleiotropy. In Drosophila, the gene that controls its wing size also affects the wing balancer, the structure of reproductive organs, fertility and lifespan of the insect. It is one gene that is the gene that is controlling the wing size that is affecting all these other characters as well. Second example is in humans. Alleles that are responsible for sickle cell anemia. What is sickle cell anemia? It is sickling of the RBC's, that the erythrocytes. It can also cause physical weakness, below average body development, hypertrophy of the bone marrow and resistance to a type of malaria. So, a person with sickle cell anemia can have these various symptoms as well. The next example is Phenylketonuria which is also known as the PKU disorder. It is a result of mutation in the phenylalanine hydroxylase gene that increases the levels of phenylalanine in the blood. The gene also causes short stature of the individual, mental deficiency, widely spaced incisors and pigmented skin and non-pigmented hair.

Now let us come to the second concept, that is penetrance. What is penetrance? Penetrance is frequency of expression of a particular phenotype among individuals carrying the corresponding genotype. So, penetrance can be of two types- complete or incomplete. Incomplete penetrance is also called as reduced penetrance. What is complete penetrance? All the individuals express the corresponding phenotype in case of complete penetrance. It means that if a person is carrying that gene and if that gene is completely penetrant, that means all the individuals carrying that gene will express that particular phenotype. The second type is the incomplete or reduced penetrance. What does it mean? It means that some individuals may fail to provide complete phenotypic expression for a genotype. That means, although the gene is there, the gene may not always express and therefore the phenotypic expression may not be complete. That's why it is incomplete. As you can see, it is depicted in these images. This is complete penetrance when in all of their all the individuals are expressing the phenotype, whereas this is incomplete penetrance wherein some of the individuals are expressing while some others are not expressing the phenotype. So, if 70% of the individuals display the phenotypic characters for a gene it inherits, the penetrance is said to be 70%. In this case you can see out of 10 individuals, 7 have expressed and three have not expressed, so here the penetrance is 70% and in complete penetrance it is always 100%. For accurate measurement of penetrance, it is necessary to consider a large population, that gives you a better idea about the penetrance of the gene. Now, some examples of complete penetrance. In garden pea, the alleles capital 'P' capital 'P' for purple flower and alleles small 'p' small 'p' for white flower have complete penetrance in the homozygous condition. It means that every time the

gene is dominant, it will express and every time it is recessive, it will not express. Let us see an example of incomplete penetrance. The polydactyly in humans is a perfect example of incomplete penetrance. It is caused by the dominant gene P. In normal condition, five digits on each limb is produced by the recessive genotype, that is, small ‘p’ small ‘p’. This gene has low penetrance. Therefore, some heterozygous individuals, that is, capital ‘P’ small ‘p’ may not be polydactylous, although there is a capital ‘P’, so that means it is incomplete penetrance. Some more examples of incomplete penetrance are the diabetes mellitus gene in humans. There may be many people carrying this gene, but it does not always cause the disease. The next example is cancer susceptibility in humans is controlled by certain genes. However, not everyone carrying the gene would develop the condition, that is, cancer. So, incomplete penetrance could be due to the presence of certain modifiers, epistatic genes or suppressors in the rest of the genome. Modifying effect of the environment also plays an important role in controlling penetrance.

Now, next concept is expressivity. Expressivity is related to penetrance, that is, a penetrant trait may exhibit variability in its phenotypic expression and this variability can be measured and is called as expressivity. Expressivity is a measure of the phenotypic variation in a population for a particular genotype. You can see here, in this population it is constant expressivity, whereas, in the second population here you can see the expressivity is varying, so this is variable expressivity. This is an example of variable expressivity. Although this is polydactylous condition there is variation in the way the gene is expressing. So, in the example of variable expressivity that is, polydactyly condition, individuals with the same genotype may have different number of digits or they may express it differently.

What is the effect of environment on penetrance or expressivity? Environmental factors such as temperature, moisture, nutrition, etc under which the organism develops has a definite effect on the penetrance and expressivity of the gene. These factors induce changes in the gene expression thereby switching the gene on and off, thereby affecting the penetrance and expressivity.

These are the books that have referred to for this module, these are the web links and this is for further reading. Thank you.