

Quadrant II – Transcript and Related Materials

Programme: Bachelor of Science (Third Year)

Subject: Botany

Course Code: BOC 108

Course Title: Cytogenetics and Plant Breeding

Unit: Mendelian genetics and its extension

Module Name: Pleiotropy, Penetrance and Expressivity

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Notes

Pleiotropy

Pleiotropy refers to the influence of a single gene on more than one phenotypic trait in an individual. The gene responsible for such an effect is called a pleiotropic gene and the effects so produced are called pleiotropic effects.

The phenomenon of pleiotropism was first observed by Gregor Mendel in his garden pea plant experiments. He saw that a single gene controlled traits such as colour of flower, colour of seed coat and red spots in leaf axils.

Some examples of pleiotropy:

1. In *Drosophila* the genes that control its wing size also affect the wing balancer, structure of reproductive organs, its fertility and also its life span.
2. In humans, alleles responsible for sickle-cell anemia (sickling of erythrocytes) also cause physical weakness, below average body development, hypertrophy of bone marrow and resistance to a type of malaria.

3. In humans, Phenylketonuria (PKU) disorder is a result of mutation in phenylalanine hydroxylase gene that increases the levels of phenylalanine in the blood. The gene also causes short stature, mental deficiency, widely spaced incisors, pigmented skin and non-pigmented hair.

Penetrance

Presence of a particular gene in an individual does not necessarily mean the gene will always express. Penetrance is the frequency of expression of a particular phenotype among individuals carrying the corresponding genotype. For accurate measurements of penetrance, it is necessary to consider a large population.

Depending on the frequency of expression, penetrance can be of two types- complete and incomplete. When all the individuals in a population carrying a particular gene express the corresponding phenotype, the gene is said to have 'complete penetrance'. However, in some cases when only certain percentage of the individuals in a population express the phenotype although all of them carry the genotype, the gene is said to have 'incomplete penetrance'.

For example, if 70% of the individuals in a population display the phenotypic character for a certain gene it inherits, the penetrance is said to be 70%.

Incomplete penetrance could be due to presence of modifiers, epistatic genes, or suppressors in the rest of the genome. Many a times it could be due to modifying effect of the environment.

Example of complete penetrance-

1. In garden pea, the alleles (PP) for purple flowers and the alleles (pp) for white flowers have complete penetrance in homozygous condition.

Example of incomplete penetrance-

1. In human, polydactyly (extra digits) is produced by dominant gene P while the normal condition of 5 digits on each limb is produced by the recessive genotype (pp). This gene has low penetrance therefore some heterozygous individuals (Pp) are not polydactylous.
2. Presence of diabetes mellitus gene in humans does not always cause the disease.
3. Cancer susceptibility in humans is controlled by certain genes. However, not everyone carrying the gene develops the condition.

Expressivity

A penetrant trait may exhibit variability in its phenotypic expression. The measure of this phenotypic variation in a population for a particular genotype is called as expressivity.

When there is no variability observed in the expressed phenotypes, the expressivity is referred to as 'constant expressivity' while when there is some degree of variability in the expressed phenotypes the expressivity is referred to as 'variable expressivity'.

For example, polydactyly demonstrates variable expressivity because individuals with the same genotype may have different numbers of digits.

Effect of environment on penetrance and 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' or 'off'.
