

Biology 1000 – Principles of Biology (Bios 115 Core II: Genetics) Exam Study Guide

1. Explain the concepts of penetrance and expressivity

Penetrance is the percentage of cases in which a certain phenotype is expressed. There are two types:

- Complete Penetrance: when 100% is correlated to the genotype. So, all express the same thing.
- Incomplete: this is when some are identical, and others are different.



$$\text{Penetrance} = \frac{\# \text{ with genotype showing phenotype}}{\# \text{ with the genotype}} \times 100$$

Expressivity is the type or degree to which a penetrant gene (the genes carrying the mutation) or genotype are phenotypically expressed.

- Constant
- Depends on the environment

Examples: Neurofibromatosis

- 50-80% penetrance (autosomal dominance)
- Variable expressivity

2. Review discussion about Breast Cancer

Breast Cancer (BRCA1 and BRCA2)

- Provides instructions for making a protein that acts as a tumor suppressor
- Prevents cells from growing and dividing
- Accounts for 30-40% of families with breast cancer cases

Penetrance and BRCA

- Refers to the risk of developing breast and ovarian cancer
- A reliable estimate of penetrance is crucial for medical counseling

Penn Risk II Model

- Predicts the probability that a person has the BRCA 1 or 2 mutant gene.
- Doesn't predict the risk of having breast cancer

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