

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:

- I. Complete Penetrance: when 100% is correlated to the genotype. So, all express the same thing.
- II. 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.

- iii. Constant
- iv. Depends on the environment

Examples: Neurofibromatosis

- a. 50-80% penetrance (autosomal dominance)
- b. Variable expressivity

2. Review discussion about Breast Cancer

Breast Cancer (BRCA1 and BRCA2)

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

Penetrance and BRCA

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

Penn Risk II Model

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

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- Chapter 1: Introduction to Biology 1000 Exam 6 Study Guide
- Chapter 2: Essential Elements of Biology 1000 Exam 6 Study Guide
- Chapter 3: Biology 1000 Exam 6 Study Guide in Everyday Life
- Chapter 4: Biology 1000 Exam 6 Study Guide in Specific Contexts
- Chapter 5: Conclusion

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This book is crafted in an easy-to-understand language and is complemented by engaging illustrations. This book is highly recommended for anyone seeking to gain a comprehensive understanding of Biology 1000 Exam 6 Study Guide.

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