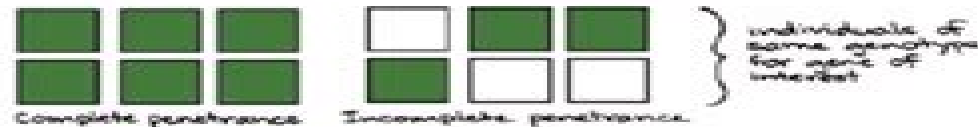


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1. Explain the concepts of penetrance and expressivity

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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**Paul Howe, Stuart Dobson, Heath
Malcolm, World Health Organization**

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Study Guide to Accompany Biology, the Science of Life, Third Edition W. J. Leverich, 1991

2,4,6-tribromophenol and Other Simple Brominated Phenols Paul Howe, Stuart Dobson, Heath Malcolm, World Health Organization, 2005-12-23 Published under the joint sponsorship of the United Nations Environment Programme the International Labour Organization and the World Health Organization and produced within the framework of the Inter organization Programme for the Sound Management of Chemicals IOMC On cover IPCS International Programme on Chemical Safety **Whitaker's Cumulative Book List**, 1961 **Life, Vol. II: Evolution, Diversity and Ecology** David E. Sadava, 2008 Co published by Sinauer Associates Inc and W H Freeman and Company Visit the Life Eighth Edition preview site LIFE HAS EVOLVED from its original publication to this dramatically revitalized Eighth Edition LIFE has always shown students how biology works offering an engaging and coherent presentation of the fundamentals of biology by describing the landmark experiments that revealed them This edition builds on those strengths and introduces several innovations As with previous editions the Eighth Edition will also be available in three paperback volumes Volume I The Cell and Heredity Chapters 1 20 Volume II Evolution Diversity and Ecology Chapters 1 21 33 52 57 Volume III Plants and Animals Chapters 1 34 51 September 2019 Monthly Current Affairs with MCQs for Competitive Exams Disha Experts, 2019-09-01

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