- c. As the proband is heterozygous for the deformity allele, the chance that the child inherited it is 1/2. If the child is also a heterozygote, the likelihood that he or she would express the defect is ~83%. Thus, the chance that the child would be affected by the deformity is 1/2 × 83% ≈ 42%.
- d. Four people in the pedigree diagram (III-1, III-2, III-4, and III-7) might have the mutant allele (because their parents had the mutant allele), but no information in the pedigree allows us to know one way or the other. If any of them do have the mutant allele, then the penetrance is lower than 83%. For example, if we somehow determined that all of those four people had the mutant allele, then the penetrance would be 5/10 = 50%, and the answer to part (c) would be 1/2 × 50% = 25%. This would be the lowest possible likelihood consistent with the data given.
- 46. a. The genotypic ratio would be 9 purple (A = B -): 7 white (A bb, aa B -, aa bb).
  - b. If the penetrance of the purple phenotype in A=B= plants is 75%, then 25% of the (A=B=) progeny would be white, and only 75% of them would be purple. This means that the purple plants in the 9/16 class would be 9/16 × 3/4 = 27/64 of the total progeny. All the remaining F<sub>2</sub> progeny [(64/64) (27/64) = 37/64] will be white. Therefore, the genotypic ratio would be 27 purple: 37 white.
  - c. If you crossed two different pure-breeding white strains, and some but not all the F<sub>1</sub> were purple, one possible explanation is incomplete penetrance of the purple phenotype. In addition, you would never be able to make a purebreeding purple strain, because even in an AA BB strain, in every generation, not all the plants will be purple.
- 47. a. Two different phenotypes are mentioned in this problem. One phenotype is the shape of the crythrocytes. All people with the genotype SPH SPH have spherical crythrocytes. Therefore, this phenotype is fully penetrant and shows no variation in expression. The second phenotype is anemia. The expressivity among anemic patients varies from severe to mild. In fact, some people with the SPH SPH genotype (150/2400) have no symptoms of anemia at all. Thus, the penetrance of the anemic phenotype is 2250/2400 or 0.94.
  - b. The severity of the anemia is greatly reduced when the spleen functions poorly and does not recognize the spherical erythrocytes as defective cells that must be eliminated from the bloodstream. Therefore, treatment might involve removing the spleen (an organ which is not essential to survival). The more efficiently the spleen functions the earlier in a patient's life it should be removed. Note that SPH SPH with no symptoms of anemia should not be subjected to this drastic treatment.
- 48. a. The most likely mode of inheritance is a single gene with incomplete dominance such that F<sup>R</sup>F<sup>R</sup> = normal (<250 mg/dl), F<sup>R</sup>F<sup>R</sup> = intermediate levels of serum cholesterol (250-500 mg/dl) and F<sup>R</sup>F<sup>R</sup> homozygotes = elevated levels (>500

# **Hartwell Solutions Manual**

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#### **Table of Contents Hartwell Solutions Manual**

- 1. Understanding the eBook Hartwell Solutions Manual
  - The Rise of Digital Reading Hartwell Solutions Manual
  - Advantages of eBooks Over Traditional Books
- 2. Identifying Hartwell Solutions Manual
  - Exploring Different Genres
  - Considering Fiction vs. Non-Fiction
  - Determining Your Reading Goals
- 3. Choosing the Right eBook Platform
  - Popular eBook Platforms
  - Features to Look for in an Hartwell Solutions Manual
  - User-Friendly Interface
- 4. Exploring eBook Recommendations from Hartwell Solutions Manual
  - Personalized Recommendations
  - Hartwell Solutions Manual User Reviews and Ratings
  - Hartwell Solutions Manual and Bestseller Lists
- 5. Accessing Hartwell Solutions Manual Free and Paid eBooks
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  - Hartwell Solutions Manual eBook Subscription Services
  - Hartwell Solutions Manual Budget-Friendly Options

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  - o ePub, PDF, MOBI, and More
  - Hartwell Solutions Manual Compatibility with Devices
  - Hartwell Solutions Manual Enhanced eBook Features
- 7. Enhancing Your Reading Experience
  - Adjustable Fonts and Text Sizes of Hartwell Solutions Manual
  - Highlighting and Note-Taking Hartwell Solutions Manual
  - Interactive Elements Hartwell Solutions Manual
- 8. Staying Engaged with Hartwell Solutions Manual
  - Joining Online Reading Communities
  - Participating in Virtual Book Clubs
  - Following Authors and Publishers Hartwell Solutions Manual
- 9. Balancing eBooks and Physical Books Hartwell Solutions Manual
  - Benefits of a Digital Library
  - Creating a Diverse Reading Collection Hartwell Solutions Manual
- 10. Overcoming Reading Challenges
  - Dealing with Digital Eye Strain
  - Minimizing Distractions
  - Managing Screen Time
- 11. Cultivating a Reading Routine Hartwell Solutions Manual
  - Setting Reading Goals Hartwell Solutions Manual
  - Carving Out Dedicated Reading Time
- 12. Sourcing Reliable Information of Hartwell Solutions Manual
  - Fact-Checking eBook Content of Hartwell Solutions Manual
  - Distinguishing Credible Sources
- 13. Promoting Lifelong Learning
  - Utilizing eBooks for Skill Development
  - Exploring Educational eBooks
- 14. Embracing eBook Trends
  - Integration of Multimedia Elements

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