

NURSING MSN 570 Advanced Pathophysiology STUDY GUIDE REVIEW EXAM 2022

WEEK 1: CELLULAR BIOLOGY AND ALTERATIONS

Week 1 Instructor Comment:

Retinoblastoma

- Retinoblastoma is a rare cancer that occurs only in children. It develops either as a hereditary disease, due to an abnormality of the RB1 gene, or sporadically (70% of cases). Located on chromosome 13q14, the RB1 gene is responsible for encoding pRb, which is a tumor suppressor protein. In the heritable form, the RB1 gene mutation is inherited through germinal cells, with a second mutation occurring in somatic retinal cells. The noninherited type of retinoblastoma occurs as a result of two mutations in the somatic retinal cells. The tumor arises from the inner surface of the retina and then spreads into the retina, resulting in leukocoria—a white appearance to the red reflex, commonly called cat-eye reflex—which is most often first identified by the child's parents.
- Normally the RB1 gene (pRB) governs the passage of the restriction point in the cell cycle, thus regulates cell proliferation.
Germline mutations are inherited, whereas somatic mutations are acquired.
- Tumor suppressors are a class of genes that work to limit cell growth, and they are important for tumor suppression by preventing DNA synthesis until mutations are corrected; preventing cell division if DNA replication introduces new mutations, and inducing programmed cell death when damage cannot be repaired. RB or retinoblastoma protein is one example of a tumor suppressor. They act in the G1/S checkpoint, thereby inhibiting progression to DNA synthesis. RB activity is regulated by many signals, including growth factors and cell differentiation. Clinically, retinoblastoma appears in the first years of life--shining light on the eye gives off an abnormal white reflection. Bilateral retinoblastoma is always due to inherited RB mutations.

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Msn Advanced Pathophysiology Study Guide

Marcel A. Müller



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Table of Contents Msn Advanced Pathophysiology Study Guide

1. Understanding the eBook Msn Advanced Pathophysiology Study Guide
 - The Rise of Digital Reading Msn Advanced Pathophysiology Study Guide
 - Advantages of eBooks Over Traditional Books
2. Identifying Msn Advanced Pathophysiology Study Guide
 - 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 Msn Advanced Pathophysiology Study Guide
 - User-Friendly Interface
4. Exploring eBook Recommendations from Msn Advanced Pathophysiology Study Guide
 - Personalized Recommendations
 - Msn Advanced Pathophysiology Study Guide User Reviews and Ratings
 - Msn Advanced Pathophysiology Study Guide and Bestseller Lists
5. Accessing Msn Advanced Pathophysiology Study Guide Free and Paid eBooks
 - Msn Advanced Pathophysiology Study Guide Public Domain eBooks
 - Msn Advanced Pathophysiology Study Guide eBook Subscription Services
 - Msn Advanced Pathophysiology Study Guide Budget-Friendly Options

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

- Interactive and Gamified eBooks

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