

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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