

Porth's Essentials of Pathophysiology 5th Edition: Chapter 5

What are congenital defects? - ANS Abnormalities of a body structure, function, or metabolism that are present at birth.

What is the leading cause of infant death? - ANS congenital defects.

The location for each gene on a chromosome is called? - ANS Locus.

A pair of loci (one from each parent) is called? - ANS Alleles.

What is reduced gene penetrance (colloquially called generation-skipping)? - ANS When an individual has the genotype to express a defect but is unaffected.

What does the basic biological anomaly in Marfan syndrome affect? - ANS Fibrillin 1, a major component of microfibrils found in the extra cellular matrix.

What are the clinical manifestations of Marfan syndrome? - ANS Myopia, vertebral deformity, long limbs and fingers, aortic aneurysms and floppy valves, and joint hyper mobility.

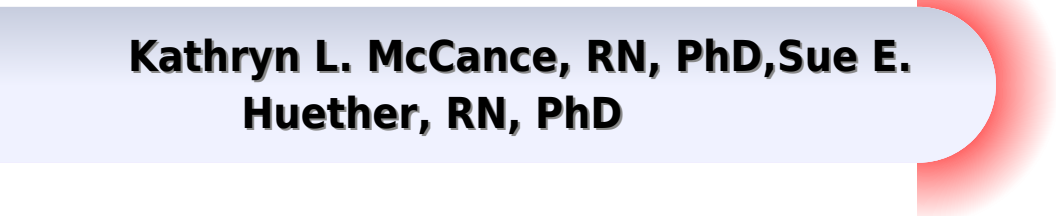
What is neurofibromatosis (NF)? - ANS Causes tumors to develop from the Schwann cells.

What are the two types of NF? - ANS Type 1 (cutaneous and subcutaneous neurofibromas that develop often in puberty. Also large, flat pigmentations [cafe au lait spots]. May also have pigmented nodules in iris) NF-1 more prone to neurological disorders and malignant neoplasms.

Type 2 (tumors of the acoustic nerve, causes headaches, hearing loss, and tinnitus).

Essentials Of Pathophysiology Test Bank Questions

**Kathryn L. McCance, RN, PhD, Sue E.
Huether, RN, PhD**



Essentials Of Pathophysiology Test Bank Questions:

Essentials Of Pathophysiology Test Bank Questions Book Review: Unveiling the Power of Words

In some sort of driven by information and connectivity, the ability of words has become more evident than ever. They have the ability to inspire, provoke, and ignite change. Such could be the essence of the book **Essentials Of Pathophysiology Test Bank Questions**, a literary masterpiece that delves deep in to the significance of words and their impact on our lives. Written by a renowned author, this captivating work takes readers on a transformative journey, unraveling the secrets and potential behind every word. In this review, we shall explore the book is key themes, examine its writing style, and analyze its overall impact on readers.

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