

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.

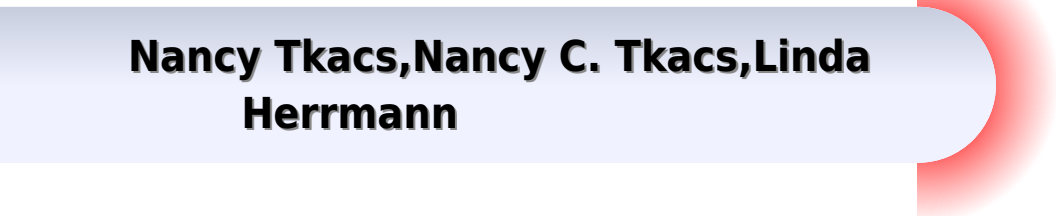
What is neurofibromatosis (NF)? - ANS Causes tumors to develop from the Schwann cells.

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

**Nancy Tkacs, Nancy C. Tkacs, Linda
Herrmann**



Essentials Of Pathophysiology Test Bank Questions:

Whispering the Techniques of Language: An Mental Quest through **Essentials Of Pathophysiology Test Bank Questions**

In a digitally-driven earth where displays reign supreme and immediate transmission drowns out the subtleties of language, the profound techniques and mental subtleties concealed within phrases often move unheard. However, set within the pages of **Essentials Of Pathophysiology Test Bank Questions** a charming literary value pulsating with natural emotions, lies an exceptional quest waiting to be undertaken. Published by a skilled wordsmith, this enchanting opus attracts readers on an introspective trip, gently unraveling the veiled truths and profound influence resonating within the very cloth of each and every word. Within the mental depths of this emotional review, we shall embark upon a heartfelt exploration of the book is core styles, dissect its charming publishing fashion, and succumb to the effective resonance it evokes strong within the recesses of readers hearts.

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