

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).

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Guillaume Favre



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