HG EXCELLENT WORK! - USE AS 1 of 2
GUIDE FOR MARCH 31, 2009

AIM: What happens when defects arise due to Spontaneous or heterogeneous lesions of DNA? (10)

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Thank you

Inherited dysfunction of a somatic change—Mutation in somatic cells is passed down to offspring due to undetected change in gametes as well

- Mutations acquired from excess radiation
- Acquired mutation in somatic cells is passed onto offspring

Haploinsufficiency—When loss of half of normal activity of a protein causes disease

- Marfan syndrome
- Shown to occur with mutations in genes encoding certain transcription factors, structural proteins, and cell surface receptors

Dominant negative—Abnormal protein synthesized, causes abnormal phenotype by interfering with function of product of normal allele

- Osteogenesis imperfecta
- Abnormal phenotype in heterozygote individuals with one copy of normal allele

Dominant gain of function—Mutant protein is enhanced in one or more of its normal properties through mutation or becomes toxic to cell through acquisition of a novel property

- Achondroplasia
- Increase in severity of disease in family history

Microdeletion—Deletion of multiple genes at closely linked loci

- Y chromosome microdeletion
- Can appear to initially follow mendelian single-gene inheritance patterns

Age of onset—Age at which a genetic disease manifests itself in the affected person.

- Huntington disease
- Congenital or late onset in affected individuals

Locus heterogeneity—The production of identical phenotypes by mutations at two or more different loci

ADD: ALLELIC HETEROGENEITY - DIFF MUTATIONS @ SAME LOCUS

- Retinitis pigmentosa
- More than one inheritance pattern correctly identified for same disease

Loss of heterozygosity—Loss of a normal allele from a region of one chromosome of a pair, allowing a defective allele on the homologous chromosome to be clinically manifest

- Retinoblastoma
- Feature of many cases of tumors due to mutation in a tumor-suppressor gene