Opening Declaration: I promise to do this assignment authentically

Student Signature: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Date:\_\_\_\_\_\_\_\_\_

1) Inherited dysfunction due to somatic change

Inherited dysfunction due to somatic change is a mutation that occurs in a precursor cell after fertilization. Any cells that come from this precursor cell will carry this mutation. This mutation may even pass onto gametes. An example of this would be a freshly fertilized zygote does not have the mutation that causes hemophilia. When the zygote undergoes its first mitotic division, one cell does not have the mutation for hemophilia, but by chance, the second daughter cell has mutated to have hemophilia. Any cells that divide from the second daughter cell will have hemophilia.

2) Haploinsufficiency

A reason for a genetic disease that involves a normal allele not being enough to cover for a dysfunctional mutated allele. An example of this is polydactyly, an autosomal dominant genetic disease. The normal allele is recessive, but the mutated allele is dominant. The fact that the normal allele is recessive means that it cannot cover for the broken function of the mutant allele.

3) Dominant negative

A dominant negative allele is an allele that causes disease by disrupting the function of the normal allele in the same cell. This can be in the form of producing a misfolded protein that bonds with normal proteins, thus causing an overall dysfunction protein complex. An example of this is with osteogensis imperfecta. The collagen molecule is composed of proα1 chains and a proα2 chain. If an allele that codes for proα1 or proα2 is mutated, then the collagen molecule as a whole becomes dysfunctional.

4)Dominant gain of function

A mutation that causes increased levels of gene activity [1]. For example, the spiked-head phenotype is caused by a point mutation in dom-1 that prevents the negative regulation of it.

5)Microdeletion

A microdeletion is a deletion that is too small to be seen under a microscope. This may result in contiguous gene syndrome where the deletion stretches between two nearby loci. For example, Miller-Dieker syndrome is caused by a microdeletion on chromosome 17. This leads to numerous problems such as congenital defects.

6)Age of onset

The age of onset is the approximate age where the symptoms begin manifesting. For example, familial adenomatous polyposis is a late onset disease that does not affect young children but affects people in their late teens.

7) Locus heterogeneity

Locus heterogeneity occurs when a mutation in at least two places produce the same effect. For example, cystic fibrosis has numerous variations for the bases that make up its mutation, but the phenotype is still the expression of cystic fibrosis.

8)Allelic heterogeneity

Allelic heterogeneity occurs when numerous different mutant alleles cause similar or even the same phenotype. An example of allelic heterogeneity is cystic fibrosis where "more than 600 different alleles can cause the associated symptoms." [2]

9) Loss of heterozygosity

A loss of heterozygosity occurs when a normal allele from one chromosome out of two is lost, causing a defective allele on the homologous chromosome to be expressed. An example of a loss of heterozygosity is with tumors that manifest in retinoblastma due to an affected tumor-supressor gene.

10)Inbreeding

Inbreeding is the act of closely related people who have children with each other, which increases the chance of recessive alleles to manifest. An example of inbreeding would be a son and a daughter from the same parents being separated at birth. Both of these kids are carriers of cystic fibrosis. If they have kids, their kids will most likely have cystic fibrosis whereas if they married normal people, the cystic fibrosis mutant allele would have been bred out.

11)Consanguinity

Consanguinity is a description of how much two people are related to each other by a common ancestor. This does not necessarily mean that these two people are closely related. For example, two people could only have a common ancestor by their great-great-great-great-grandfather. They would be distantly related.

12) Sex influence

Sex influence refers to how certain traits that are not X-linked are expressed differently in males and females. An example is how facial hair on men tends to be darker and more coarse in comparison to females that have lighter facial hair.

Bibliography

1. Fay, David, and Andy Spencer. "Genetic Mapping and Manipulation." Wormbook. N.p., 17 Feb. 2006. Web. 07 Apr. 2016. <http://www.wormbook.org/chapters/www\_dominantmu tations/dominantmutations.html#d0e484>.

2. "Defining Mendelian Genetics and Underlying Concepts." Genetics and Nutrition. American Dietetic Association and National Genetics Education and Development Centre, 2016. Web. 07 Apr. 2016 <http://www.nchpeg.org/nutrition/index.php?option=com\_content& view=article&id=406>.

Closing Declaration: At the close of this assignment, I can attest to having done it by my own hand. If I received help from peers or from tutors in doing it, this was purely to understand the material, and I did not knowingly transfer the information from or to other sources (my peers or otherwise) in the process of doing this work

Student Signature: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Date:\_\_\_\_\_\_\_\_\_