Mutations, Dominance, and Haplosufficiency

Throughout the genetics course, you will learn about many types of mutations and what they mean for a phenotype of an individual. The concepts of mutations, dominance, and haplosufficiency will act as a foundation to build the rest of the concepts for this course.

Mutations

DNA is the genetic material that codes for the production of proteins. A change in the DNA of an organism can be called a mutation. Mutations can lead to a change in phenotype. This change of phenotype can be a visual change or something someone could measure (such as the amount of an enzyme produced in the body or a blood type).

Not all mutations are bad; many traits seen in living humans today came from mutations. Two common misconceptions for this course surround mutations and their phenotypes. A mutation can be dominant or recessive. The mutant phenotype can be something that helps an organism survive or hinders them. This means mutations can be good; they are not all bad. In addition to this, not all alleles that are expressed as a detrimental phenotype are considered mutations. Do not call an allele a mutant allele unless your professor provides a clue that it is a mutation.

Alleles

A diploid individual has two copies of every gene, but these two copies (alleles) can be slightly different. Each allele codes for a variant of a characteristic. Fur color can act as a good example to think about alleles. There is a gene for fur color. The variants of fur color (brown, red, black, white, or orange) are coded for by different alleles. A diploid individual could be heterozygous meaning they have two different alleles or have two of the same alleles (homozygous recessive or homozygous dominant).

Dominance In Respect to Alleles

Dominance refers to which allele will be expressed in a phenotype. A dominant allele will always be expressed regardless of if there is a recessive allele also. This means that heterozygous individuals will always express the dominant allele and have a dominant phenotype. An individual who is homozygous recessive (has two recessive alleles) will express the recessive phenotype and an individual who is homozygous dominant (has two dominant alleles) will express the dominant phenotype.

Haplosufficiency

Haplosufficiency is a topic which brings together the topics of mutations and dominance. A wildtype allele is the original allele in a population. Mutations to DNA can result in other alleles, some of which may be referred to as mutant alleles.

A gene is considered to be haplosufficient if only one wildtype allele is required to show a wildtype phenotype. This means that the wildtype allele is dominant when the gene is haplosufficient. However, not all wildtype alleles are dominant. When a wildtype allele is recessive to a mutant allele, the gene is considered haploinsufficient. This means that one wildtype allele in a genotype is not enough for an individual to show/ express a wildtype phenotype.

Learning Goals:

- Comprehend that a mutation can result in phenotypes that are beneficial or detrimental to an organism.
- Understand dominance and be able to explain its relation to alleles, the expression of alleles, genotypes, and phenotypes.
- Understand the definition of haplosufficiency.
- Be able to indicate if a gene is haplosufficient or haploinsufficient based on an individual's genotype and phenotype or knowing which allele is dominant.

Order of Activities:

1. Review what dominance means by reading this short summary: https://www.genome.gov/genetics-glossary/Dominant
2. Read the following summary sheet about haplosufficiency.

3. Test yourself by completing the corresponding worksheet for this material. Attempt to first complete this on your own, then pair up with a partner or group to discuss when possible. There is an answer key provided so you can check your work and read explanations of how to answer the problem. Any questions you get wrong or confused about you should attempt to explain why the answer is correct and then complete again after you finish the activities in this guide.
4. After reviewing any topic, it is a good idea to have a metacognition check. Ask yourself the following questions:

    - What are my emotional responses to learning this material? Which material am I frustrated with and need aid in understanding?
    - What difficulties have I had with the learning tasks? What specific tasks will I do to master this content?
    - Do I understand all of the learning goals? Can I explain each of them out loud to someone clearly and concisely?
    - How is what I learned related to other things I have learned in this class? How is it related to other classes, my career, and my life?

5. If you would like to have more aid in learning this material, please reach out. There are numerous individuals who want to help you feel confident in your understanding. If your course has learning assistants or teaching assistant(s), you should reach out to them to review concepts you want to learn more about. Your professor is also a great resource to go to when you do not understand a topic. You can study with your peers or receive academic support through the LRC as well. If you would like help identifying how to receive the support you need, do not hesitate to contact the CU Denver Learning Resources Center at LRC@ucdenver.edu or stop by our front desk in the learning commons building.