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1 | SNP. Project. Write-up.

Resources: [[KBxSNPPCR]] Instructions

1.0.1 | Part One

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Part 1: Gene and associated SNP description:

This introductory section will help the reader understand what is known about your gene/SNP: SNP context within the gene, typical gene/protein function and regulation, possible human genotypes at the SNP, connections to phenotypes, etc. In Part 2, you will provide an infographic companion to this section. This section should be written in your own words, but based on at least three primary or review article sources from the scientific literature in addition to several databases (linked below). You should not rely on SNPedia write-ups as your source material; dig into the literature yourself. Cite your sources in-line using superscript numbers or parenthetical APA style.

Include the following elements in the order/format that makes most sense to you, along with any other information you deem relevant:

- Gene/SNP Basics - you may integrate the following basic information within your text as you describe gene function and regulation. This doesn’t need to be a set-apart section. General gene info can be found in the NCBI Gene database. SNP info can be found at dbSNP by searching with your SNP rs#.
- Full gene name
- Gene abbreviation (capitalized and italicized)
- Protein name (capitalized, non-italicized; usually the same as the gene abbreviation)
- Chromosome #
- Position of SNP within the gene (protein-coding or non-protein coding, regulatory region, etc
- SNP allele frequencies in the global human population (for both the major and minor allele(s)). For example, G = 79%; T = 21%. You can use the total population allele frequencies reported by ALFA in the dbSNP frequency tab for your specific SNP. Example here for APOE SNP rs429358.
- Note that allele frequencies aren’t the same as genotype frequencies (expected genotype frequencies from allele frequencies above of G 79% and T 21%: G-G = 62.4%; G-T = 33.2%; T-T = 4.4%). Allele frequencies can be used to calculate expected genotype frequencies using the assumptions of the Hardy-Weinberg equation. You don’t have to do this for your write-up but you can if you’re interested.
- Gene function and regulation -
- Explain what the wild-type (the typical or “normal”) gene and protein do at the cellular and organismal level. What is known about this protein’s typical function?

- You can use gene/protein databases and/or published research reports to learn about your protein's typical function. Examples for ACE2 protein: NCBI gene report; UniProt report; google scholar on ACE2 function.
- Where is the gene expressed in the body and in (or outside of) cells (which gives you an idea of where the protein performs its function)? What regulates the expression of this gene: are there known transcription factor(s) or environmental signals (chemicals, dietary substances, etc)?
- To understand tissue expression patterns, look at the NCBI gene entry for your gene and scroll down to the "expression" section. For a bit more information, check out your gene in the Human Protein Atlas (example showing ACTL6B with high brain expression in the nucleus of particular neurons).
- SNP connections to function and phenotypes - How are the variants/genotypes at your SNP position connected to gene function and/or human phenotypes? Dig into the nuances here; there may not be a super strong correlation between genotypes and phenotypes because of the polygenic nature of inheritance and the influence of the environment on our phenotypes/traits. That's okay, and you can acknowledge it in your analysis. You may use the information about the wild-type version of the gene as a point of comparison for any SNP variants that have known alternative phenotypes.

Search the published scientific literature with google scholar or PubMed and/or use links to published articles that you've found in SNPedia or dbSNP databases.**