# L13 SIRE506 Homework

Deadline: 08-04-2019

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## Question 1:

For each of these variants …:

* rs13447331
* rs16904774
* rs13398235

Locate the following information:

* Allele frequency of alternative allele(s). Use the allele frequency for the population most relevant to someone from Thailand.
* Location (if in a gene, provide gene name and which exon/intron).
* Predicted impact on phenotype.

## Question 2:

What is “compound heterozygous inheritance” and why is it important? Explain how to determine if two heterozygote variants are from compound heterozygous inheritance.

## Question 3:

What does a “stop retained” variant refers to and what impact does it have on the gene?

## Question 4:

Explain why these variants might not have an impact on the phenotype

* “stop\_gained” towards the end of the gene.
* “frameshift\_variant” in the second of ten exons.

Give an example of a situation where an “intergenic\_variant” might be pathogenic.

## Question 5:

How and why can conservation of gene sequence be used to predict whether variants are more likely to be pathogenic or benign?

## Question 6:

Use the provided clinical symptoms and the VCF file to answer the following questions (explain your reasoning):

What is the most likely disease(s) based only on the provided symptoms?

Which variant is most likely the causative mutation? Does this fit with your answer for the previous question?

Symptoms:

* Aminoaciduria
* Abnormality of cardiovascular system morphology
* Intellectual disability, severe

Variants:

* See attached VCF file