# Analyse Variants

We have analysed germline variants as part of our three clinical cases. We have also looked at a somatic variant and the somatic variant guidelines in the identify variation competency.

In the protein sequence competency, we looked at how some of the bioinformatics prediction tools work to predict whether a variant is potentially benign or potentially pathogenic.

## General Questions

1. What (briefly) does the laboratory genomics service do? How does this help patients?
2. What do bioinformaticians in laboratory genomics do (hint- have a look at the bioinformatics best practice guidelines)?
3. What website(s) might you use to search the published literature for information on a variant or genetic condition?

## Resources

<http://www.wales.nhs.uk/sites3/page.cfm?orgid=525&pid=19419>

<https://www.acgs.uk.com/quality/best-practice-guidelines/> (go to Sequencing Guidelines section and open the document entitled NEW! NGS Bioinformatics August 2016)