Question 2

Determine genetic basis of protein alterations

1. Sequence analysis to identify if alterations are monoallelic or biallelic
2. Identify any associations, or any anticorrelations, with other genetic alterations

Loss of heterozygosity – identify copy number values (abs – major/minor allele; if minor allele 0 then indicates only major -> LoH)

* 1. Compare to other genes in PCAWG
  2. Compare to other HR genes

Plan

* Obtain only exonic mutations of RAD51B
  + Select mutation ids
  + Identify donor ids
    - Identify how many have germline variant data available
    - Extract germline data for LoH analysis
* CNV
  + Copy number values, replot summary table
  + Histogram/bar chart of CNV change vs. (for profile comparison)
    - Other genes in PCAWG (randomly selected) [relevant?]
    - HR genes
    - Known tumour suppressors and oncogenes
* SV
  + SV summary
    - Frequency of type of SVs
    - SVs per sample
    - Mean SVs
    - Median SVs
  + Compare to
    - Other genes in PCAWG (randomly selected) [relevant?]
    - HR genes
    - Known tumour suppressors and oncogenes
* SNV/Indel
  + SNV/Indel summary
    - Frequency of type of SNVs
    - SNVs per sample
    - Mean SNVs
    - Median SNVs
  + Compare to
    - Other genes in PCAWG (randomly selected) [relevant?]
    - HR genes
    - Known tumour suppressors and oncogenes