

## Additional file 2 – Table – FunctSNP SNP prioritisation

| NCBI's Function Classification                           | Score |
|----------------------------------------------------------|-------|
| Changes to STOP codon (nonsense)                         | 20    |
| Alters codon to make an altered amino acid<br>(missense) | 20    |
| indel SNP causing frame shift                            | 20    |
| Synonymous amino acid change                             | 15    |
| Within 3' 0.5kb to a gene                                | 10    |
| Within 5' 2kb to a gene                                  | 10    |
| Untranslated region                                      | 5     |
| Protein coding: synonymy unknown                         | 5     |
| 3 prime untranslated                                     | 5     |
| 5 prime untranslated region                              | 5     |
| Slice site - 3 prime acceptor dinucleotide               | 5     |
| Slice site - 5 prime donor dinucleotide                  | 5     |
| Intron                                                   | 1     |