**Table S5:** Web resources

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| --- | --- | --- |
| **Name** | **Available** | **Accessed** |
| 1000 genomes  | <http://www.1000genomes.org/> | Nov 2012 |
| Database of Single Nucleotide Polymorphisms (dbSNP Build ID: 134) | <http://www.ncbi.nlm.nih.gov/SNP/> | Nov 2012 |
| Ensembl60 | <http://www.ensembl.org/index.html> | Nov 2012 |
| Exome Variant Server, NHLBI Exome Sequencing Project (ESP) | <http://evs.gs.washington.edu/EVS/> | Nov 2012 |
| Human Splicing Finder | <http://www.umd.be/HSF/> | Nov 2012 |
| Online Mendelian Inheritance in Man (OMIM) | <http://www.omim.org/> | Nov 2012 |
| NNsplice - **Berkeley Drosophila Genome Project** | <http://www.fruitfly.org/seq_tools/splice.html> | Nov 2012 |
| PolyPhen-2  | <http://genetics.bwh.harvard.edu/pph2/> | Nov 2012 |
| SIFT | <http://sift.jcvi.org/> | Nov 2012 |