

Supplemental material

Online resources

dbSNP: <http://www.ncbi.nlm.nih.gov/projects/SNP/>; 1000 Genomes:

<http://browser.1000genomes.org/>; Exome Aggregation Consortium [ExAC]:

<http://exac.broadinstitute.org/>; NHLBI Exome Sequencing Project Exome Variant Server

[EVS]: <http://evs.gs.washington.edu/EVS/>; PolyPhen-2:

<http://genetics.bwh.harvard.edu/pph2/>; SIFT: <http://sift.jcvi.org/>; Mutation Assessor:

<http://mutationassessor.org/>; Provean: <http://provean.jcvi.org/>; HaplotypeCaller and

GATK: <https://www.broadinstitute.org/gatk/>; Clustal Omega:

<http://www.ebi.ac.uk/Tools/msa/clustalo/>; LOVD: <http://LOVD.nl/CEP120>.