LBB-Mendelics-2021 Dia 3

**Questão 1: Obtenha a razão Ti/Tv (transitions e transversions) das variantes encontrada no cromossomo 22.**

Resposta: Foi usado 3 ferramentas (vcftools, bcftools e vcflib) para o cálculo Ti/Tv para garantir o valor correto. O valor obtido pelas ferramentas vcftools e bcftools foi de 2.91. o valor obtido pela ferramenta vcflib foi de 2.88.

# vcftools

$ cat amostra-lbb.vcftools\_filter\_step2.recode.vcf.eff | vcf-tstv

**2.91** 689 (ts=513 tv=176 total=735 skipped=46 multiallelic=0)

# bcftools

$ bcftools stats amostra-lbb.vcftools\_filter\_step2.recode.vcf.eff | grep "TSTV"

# TSTV, transitions/transversions:

# TSTV [2]id [3]ts [4]tv [5]ts/tv [6]ts (1st ALT) [7]tv (1st ALT) [8]ts/tv (1st ALT)

TSTV 0 513 176 2.91 513 176 **2.91**

# vcflib

$ vcfstats amostra-lbb.vcftools\_filter\_step2.recode.vcf

total variant sites: 735

of which 735 (1) are biallelic and 0 (0) are multiallelic

total variant alleles: 735

unique variant alleles: 737

snps: 691

mnps: 0

indels: 46

complex: 2

mismatches: 691

ts/tv ratio: **2.88202**

deamination ratio: 1.29018

biallelic snps: 689 @ 2.91477

**Questão 2: Quantas variantes são encontradas na região de 16000000 a 20000000?**

Resposta: 64 variantes

$ grep -v "#" amostra-lbb.vcftools\_filter\_step2.recode.vcf.eff | awk '{if ($2 >= 16000000 && $2 <= 20000000) print $2}' | wc -l

64

**Questão 3: Exiba o conteúdo da linha do VCF relativa a uma variante:**

**- Não-sinônima**

**- Variante no gnomAD v3.1.1 com MAF < 0.01.**

Resposta: Para a anotação das variantes foram utilizados softwares snpSift e snpEff de forma a obter os efeitos e os impacto em variantes no transcrito canônico (-lof -classic -hgvs -canon -ud 0). As bases do 1000 Genomas quanto a base dbNSFP(v4.1) foram utilizadas. Dessa forma, foram encontradas 56 variantes não-sinônimas com MAF < 0.01 após o filtro com o vcftools.

# Para executar o vcftools foi necessário apagar a linha 322 do arquivo amostra-lbb.vcftools\_filter\_step2.recode.vcf.eff, pois o parser do vcftools não estava reconhecendo a flag dbNSFP\_VEP\_canonical

Error: Flag Type must have 0 entries: ID=dbNSFP\_VEP\_canonical,Number=A,Type=Flag,Description="Field 'VEP\_canonical' from dbNSFP">

$ vcftools --vcf amostra-lbb.vcftools\_filter\_step2.recode.vcf.eff --max-maf 0.01 --recode --recode-INFO-all --out amostra-lbb.vcftools\_maf\_filter

$ grep -v "#" amostra-lbb.vcftools\_maf\_filter.recode.vcf | grep "EFF=NON\_" | wc -l

56

O conteúdo da resposta da variante pedida é de uma variante tanto não-sinônima quanto uma variante presente na base do gnomAD v3.1.1 com MAF < 0.01.

Frequência alélica da variante no gnomAD v3.1.1:

dbNSFP\_gnomAD\_exomes\_AF=4.798531e-01

Variante:

chr22:50625611-50625611

Conteúdo da linha:

chr22 50625611 . G C 4476.06 PASS AC=2;AF=1;AN=2;DP=132;ExcessHet=3.0103;FS=0;MLEAC=2;MLEAF=1;MQ=60;QD=35.32;SOR=0.808;EFF=NON\_SYNONYMOUS\_CODING(MODERATE|MISSENSE|aCt/aGt|p.Thr393Ser/c.1178C>G|509|ARSA|protein\_coding|CODING|NM\_000487.6|7|C);dbNSFP\_1000Gp3\_AC=2031;dbNSFP\_1000Gp3\_AF=0.4055511182108626;dbNSFP\_1000Gp3\_AFR\_AC=473;dbNSFP\_1000Gp3\_AFR\_AF=0.3577912254160363;dbNSFP\_1000Gp3\_AMR\_AC=326;dbNSFP\_1000Gp3\_AMR\_AF=0.4697406340057637;dbNSFP\_1000Gp3\_EAS\_AC=324;dbNSFP\_1000Gp3\_EAS\_AF=0.32142857142857145;dbNSFP\_1000Gp3\_EUR\_AC=532;dbNSFP\_1000Gp3\_EUR\_AF=0.5288270377733598;dbNSFP\_1000Gp3\_SAS\_AC=376;dbNSFP\_1000Gp3\_SAS\_AF=0.38445807770961143;dbNSFP\_ALSPAC\_AC=2095;dbNSFP\_ALSPAC\_AF=0.5435910742086144;dbNSFP\_APPRIS=principal1,principal1,principal1,.,principal1;dbNSFP\_Aloft\_Confidence=.,.,.,.,.;dbNSFP\_Aloft\_Fraction\_transcripts\_affected=.,.,.,.,.;dbNSFP\_Aloft\_pred=.,.,.,.,.;dbNSFP\_Aloft\_prob\_Dominant=.,.,.,.,.;dbNSFP\_Aloft\_prob\_Recessive=.,.,.,.,.;dbNSFP\_Aloft\_prob\_Tolerant=.,.,.,.,.;dbNSFP\_AltaiNeandertal=C/C;dbNSFP\_Ancestral\_allele=C;dbNSFP\_BayesDel\_addAF\_pred=T;dbNSFP\_BayesDel\_addAF\_rankscore=0.00612;dbNSFP\_BayesDel\_addAF\_score=-0.4948;dbNSFP\_BayesDel\_noAF\_pred=T;dbNSFP\_BayesDel\_noAF\_rankscore=0.40377;dbNSFP\_BayesDel\_noAF\_score=-0.339702;dbNSFP\_CADD\_phred=0.664;dbNSFP\_CADD\_phred\_hg19=11.23;dbNSFP\_CADD\_raw=-0.146328;dbNSFP\_CADD\_raw\_hg19=0.710054;dbNSFP\_CADD\_raw\_rankscore=0.03166;dbNSFP\_CADD\_raw\_rankscore\_hg19=0.13303;dbNSFP\_ClinPred\_pred=T;dbNSFP\_DANN\_rankscore=0.14473;dbNSFP\_DANN\_score=0.82971795878766041;dbNSFP\_DEOGEN2\_pred=T,T,T,.,T;dbNSFP\_DEOGEN2\_rankscore=0.59137;dbNSFP\_DEOGEN2\_score=0.226365,0.226365,0.226365,.,0.226365;dbNSFP\_Denisova=C/C;dbNSFP\_ESP6500\_AA\_AC=1736;dbNSFP\_ESP6500\_AA\_AF=0.3940081706763504;dbNSFP\_ESP6500\_EA\_AC=4560;dbNSFP\_ESP6500\_EA\_AF=0.5302325581395348;dbNSFP\_Eigen\_PC\_phred\_coding=0.4775234;dbNSFP\_Eigen\_PC\_raw\_coding=-1.00922207557023;dbNSFP\_Eigen\_PC\_raw\_coding\_rankscore=0.09581;dbNSFP\_Eigen\_phred\_coding=0.2570364;dbNSFP\_Eigen\_raw\_coding=-1.15937330945236;dbNSFP\_Eigen\_raw\_coding\_rankscore=0.05627;dbNSFP\_Ensembl\_geneid=ENSG00000100299,ENSG00000100299,ENSG00000100299,ENSG00000100299,ENSG00000100299;dbNSFP\_Ensembl\_proteinid=ENSP00000348406,ENSP00000216124,ENSP00000378983,ENSP00000412542,ENSP00000378981;dbNSFP\_Ensembl\_transcriptid=ENST00000356098,ENST00000216124,ENST00000395621,ENST00000453344,ENST00000395619;dbNSFP\_ExAC\_AC=58673;dbNSFP\_ExAC\_AF=4.833e-01;dbNSFP\_ExAC\_AFR\_AC=4080;dbNSFP\_ExAC\_AFR\_AF=3.941e-01;dbNSFP\_ExAC\_AMR\_AC=4804;dbNSFP\_ExAC\_AMR\_AF=4.156e-01;dbNSFP\_ExAC\_Adj\_AC=58572;dbNSFP\_ExAC\_Adj\_AF=4.843e-01;dbNSFP\_ExAC\_EAS\_AC=2570;dbNSFP\_ExAC\_EAS\_AF=2.984e-01;dbNSFP\_ExAC\_FIN\_AC=3916;dbNSFP\_ExAC\_FIN\_AF=5.923e-01;dbNSFP\_ExAC\_NFE\_AC=35803;dbNSFP\_ExAC\_NFE\_AF=5.393e-01;dbNSFP\_ExAC\_SAS\_AC=6967;dbNSFP\_ExAC\_SAS\_AF=4.220e-01;dbNSFP\_ExAC\_nonTCGA\_AC=51025;dbNSFP\_ExAC\_nonTCGA\_AF=4.804e-01;dbNSFP\_ExAC\_nonTCGA\_AFR\_AC=3535;dbNSFP\_ExAC\_nonTCGA\_AFR\_AF=3.911e-01;dbNSFP\_ExAC\_nonTCGA\_AMR\_AC=4657;dbNSFP\_ExAC\_nonTCGA\_AMR\_AF=4.154e-01;dbNSFP\_ExAC\_nonTCGA\_Adj\_AC=51021;dbNSFP\_ExAC\_nonTCGA\_Adj\_AF=4.807e-01;dbNSFP\_ExAC\_nonTCGA\_EAS\_AC=2342;dbNSFP\_ExAC\_nonTCGA\_EAS\_AF=2.989e-01;dbNSFP\_ExAC\_nonTCGA\_FIN\_AC=3916;dbNSFP\_ExAC\_nonTCGA\_FIN\_AF=5.923e-01;dbNSFP\_ExAC\_nonTCGA\_NFE\_AC=29321;dbNSFP\_ExAC\_nonTCGA\_NFE\_AF=5.396e-01;dbNSFP\_ExAC\_nonTCGA\_SAS\_AC=6918;dbNSFP\_ExAC\_nonTCGA\_SAS\_AF=4.216e-01;dbNSFP\_ExAC\_nonpsych\_AC=41898;dbNSFP\_ExAC\_nonpsych\_AF=4.618e-01;dbNSFP\_ExAC\_nonpsych\_AFR\_AC=4069;dbNSFP\_ExAC\_nonpsych\_AFR\_AF=3.937e-01;dbNSFP\_ExAC\_nonpsych\_AMR\_AC=4797;dbNSFP\_ExAC\_nonpsych\_AMR\_AF=4.154e-01;dbNSFP\_ExAC\_nonpsych\_Adj\_AC=41797;dbNSFP\_ExAC\_nonpsych\_Adj\_AF=4.628e-01;dbNSFP\_ExAC\_nonpsych\_EAS\_AC=1716;dbNSFP\_ExAC\_nonpsych\_EAS\_AF=3.049e-01;dbNSFP\_ExAC\_nonpsych\_FIN\_AC=2250;dbNSFP\_ExAC\_nonpsych\_FIN\_AF=5.921e-01;dbNSFP\_ExAC\_nonpsych\_NFE\_AC=21675;dbNSFP\_ExAC\_nonpsych\_NFE\_AF=5.186e-01;dbNSFP\_ExAC\_nonpsych\_SAS\_AC=6962;dbNSFP\_ExAC\_nonpsych\_SAS\_AF=4.219e-01;dbNSFP\_FATHMM\_converted\_rankscore=0.89363;dbNSFP\_FATHMM\_pred=D,D,D,D,D;dbNSFP\_FATHMM\_score=-2.52,-2.52,-2.52,-2.52,-2.52;dbNSFP\_GENCODE\_basic=Y,Y,Y,Y,Y;dbNSFP\_GERP\_\_\_NR=5.59;dbNSFP\_GERP\_\_\_RS=-0.0288;dbNSFP\_GERP\_\_\_RS\_rankscore=0.13247;dbNSFP\_GM12878\_confidence\_value=0;dbNSFP\_GTEx\_V8\_gene=CPT1B\_CHKB\_ARSA\_CPT1B\_CHKB-DT\_CHKB\_ARSA\_CPT1B\_CHKB\_ARSA\_CPT1B\_CHKB\_ARSA\_SYCE3\_ARSA\_CHKB\_ARSA\_CPT1B\_CHKB\_ARSA\_CPT1B\_ARSA\_CHKB\_CPT1B\_CHKB-DT\_CHKB\_ARSA\_MAPK8IP2\_CHKB\_ARSA\_CHKB\_ARSA\_ARSA\_ARSA\_CPT1B\_CHKB\_ARSA\_CPT1B\_CHKB\_CPT1B\_CHKB\_ARSA\_CPT1B\_CHKB\_ARSA\_CPT1B\_CHKB\_CPT1B\_CHKB\_ARSA\_CHKB\_ARSA\_CPT1B\_CHKB\_ARSA\_ARSA\_CPT1B\_CHKB\_ARSA\_CPT1B\_ARSA;dbNSFP\_GTEx\_V8\_tissue=Adipose\_Subcutaneous\_Adipose\_Subcutaneous\_Adipose\_Subcutaneous\_Adipose\_Visceral\_Omentum\_Adipose\_Visceral\_Omentum\_Adipose\_Visceral\_Omentum\_Adipose\_Visceral\_Omentum\_Artery\_Aorta\_Artery\_Aorta\_Artery\_Aorta\_Artery\_Tibial\_Artery\_Tibial\_Artery\_Tibial\_Brain\_Caudate\_basal\_ganglia\_Brain\_Hippocampus\_Breast\_Mammary\_Tissue\_Breast\_Mammary\_Tissue\_Cells\_Cultured\_fibroblasts\_Cells\_Cultured\_fibroblasts\_Cells\_Cultured\_fibroblasts\_Colon\_Transverse\_Colon\_Transverse\_Esophagus\_Gastroesophageal\_Junction\_Esophagus\_Mucosa\_Esophagus\_Mucosa\_Esophagus\_Mucosa\_Esophagus\_Mucosa\_Esophagus\_Muscularis\_Esophagus\_Muscularis\_Esophagus\_Muscularis\_Heart\_Atrial\_Appendage\_Heart\_Atrial\_Appendage\_Heart\_Left\_Ventricle\_Liver\_Lung\_Lung\_Lung\_Muscle\_Skeletal\_Muscle\_Skeletal\_Nerve\_Tibial\_Nerve\_Tibial\_Nerve\_Tibial\_Pancreas\_Pancreas\_Pituitary\_Skin\_Not\_Sun\_Exposed\_Suprapubic\_Skin\_Not\_Sun\_Exposed\_Suprapubic\_Skin\_Sun\_Exposed\_Lower\_leg\_Skin\_Sun\_Exposed\_Lower\_leg\_Skin\_Sun\_Exposed\_Lower\_leg\_Spleen\_Spleen\_Stomach\_Stomach\_Stomach\_Testis\_Thyroid\_Thyroid\_Thyroid\_Whole\_Blood\_Whole\_Blood;dbNSFP\_GenoCanyon\_rankscore=0.24082;dbNSFP\_GenoCanyon\_score=0.794969999019877;dbNSFP\_Geuvadis\_eQTL\_target\_gene=ENSG00000100299;dbNSFP\_H1\_hESC\_confidence\_value=0;dbNSFP\_H1\_hESC\_fitCons\_rankscore=0.48771;dbNSFP\_H1\_hESC\_fitCons\_score=0.645312;dbNSFP\_HGVSc\_VEP=c.1178C>G,c.1178C>G,c.1178C>G,c.920C>G,c.1178C>G;dbNSFP\_HGVSp\_VEP=p.Thr393Ser,p.Thr393Ser,p.Thr393Ser,p.Thr307Ser,p.Thr393Ser;dbNSFP\_HGVSp\_snpEff=p.Thr393Ser,p.Thr393Ser,p.Thr393Ser,p.Thr307Ser,p.Thr393Ser;dbNSFP\_HUVEC\_confidence\_value=0;dbNSFP\_HUVEC\_fitCons\_rankscore=0.41899;dbNSFP\_HUVEC\_fitCons\_score=0.613276;dbNSFP\_Interpro\_domain=.,.,.,.,.;dbNSFP\_LINSIGHT=.;dbNSFP\_LINSIGHT\_rankscore=.;dbNSFP\_LIST\_S2\_pred=.,.,.,T,T;dbNSFP\_LIST\_S2\_rankscore=0.00005;dbNSFP\_LIST\_S2\_score=.,.,.,0.00239976,0.00217978;dbNSFP\_LRT\_Omega=1.175200;dbNSFP\_LRT\_converted\_rankscore=0.05919;dbNSFP\_LRT\_pred=N;dbNSFP\_LRT\_score=0.650226;dbNSFP\_M\_CAP\_pred=.;dbNSFP\_M\_CAP\_rankscore=.;dbNSFP\_M\_CAP\_score=.;dbNSFP\_MPC\_rankscore=.;dbNSFP\_MPC\_score=.,.,.,.,.;dbNSFP\_MVP\_rankscore=.;dbNSFP\_MVP\_score=.,.,.,.,.;dbNSFP\_MetaLR\_pred=T;dbNSFP\_MetaLR\_rankscore=0.00011;dbNSFP\_MetaLR\_score=0.0000;dbNSFP\_MetaSVM\_pred=T;dbNSFP\_MetaSVM\_rankscore=0.24625;dbNSFP\_MetaSVM\_score=-1.0172;dbNSFP\_MutPred\_AAchange=.;dbNSFP\_MutPred\_protID=.;dbNSFP\_MutPred\_rankscore=.;dbNSFP\_MutPred\_score=.;dbNSFP\_MutationAssessor\_pred=.,.,.,.,.;dbNSFP\_MutationAssessor\_rankscore=.;dbNSFP\_MutationAssessor\_score=.,.,.,.,.;dbNSFP\_MutationTaster\_AAE=T391S,T393S,T307S,T393S,T393S,T393S,T391S;dbNSFP\_MutationTaster\_converted\_rankscore=0.08975;dbNSFP\_MutationTaster\_model=simple\_aae,simple\_aae,simple\_aae,simple\_aae,simple\_aae,simple\_aae,simple\_aae;dbNSFP\_MutationTaster\_pred=P,P,P,P,P,P,P;dbNSFP\_MutationTaster\_score=1,1,1,1,1,1,1;dbNSFP\_PROVEAN\_converted\_rankscore=0.05503;dbNSFP\_PROVEAN\_pred=N,N,N,N,N;dbNSFP\_PROVEAN\_score=0.13,0.13,0.13,0.18,0.13;dbNSFP\_Polyphen2\_HDIV\_pred=.,.,.,.,.;dbNSFP\_Polyphen2\_HDIV\_rankscore=.;dbNSFP\_Polyphen2\_HDIV\_score=.,.,.,.,.;dbNSFP\_Polyphen2\_HVAR\_pred=.,.,.,.,.;dbNSFP\_Polyphen2\_HVAR\_rankscore=.;dbNSFP\_Polyphen2\_HVAR\_score=.,.,.,.,.;dbNSFP\_PrimateAI\_pred=T;dbNSFP\_PrimateAI\_rankscore=0.19553;dbNSFP\_PrimateAI\_score=0.361151695251;dbNSFP\_REVEL\_rankscore=0.61300;dbNSFP\_REVEL\_score=0.290;dbNSFP\_Reliability\_index=9;dbNSFP\_SIFT4G\_converted\_rankscore=0.14588;dbNSFP\_SIFT4G\_pred=T,T,T,T,T;dbNSFP\_SIFT4G\_score=0.414,0.414,0.414,0.409,0.414;dbNSFP\_SIFT\_converted\_rankscore=0.08891;dbNSFP\_SIFT\_pred=T,T,T,T,T;dbNSFP\_SIFT\_score=0.469,0.469,0.469,0.454,0.469;dbNSFP\_SiPhy\_29way\_logOdds=1.5892;dbNSFP\_SiPhy\_29way\_logOdds\_rankscore=0.02491;dbNSFP\_SiPhy\_29way\_pi=0.1364:0.3411:0.1333:0.3891;dbNSFP\_TSL=1,1,2,2,5;dbNSFP\_TWINSUK\_AC=2004;dbNSFP\_TWINSUK\_AF=0.540453074433657;dbNSFP\_UK10K\_AC=4099;dbNSFP\_UK10K\_AF=0.5420523670986511;dbNSFP\_Uniprot\_acc=A0A0C4DFZ2,A0A0C4DFZ2,A0A0C4DFZ2,P15289-2,A0A0C4DFZ2;dbNSFP\_Uniprot\_entry=A0A0C4DFZ2\_HUMAN,A0A0C4DFZ2\_HUMAN,A0A0C4DFZ2\_HUMAN,ARSA\_HUMAN,A0A0C4DFZ2\_HUMAN;dbNSFP\_VEP\_canonical;dbNSFP\_VEST4\_rankscore=0.00825;dbNSFP\_VEST4\_score=0.031,0.031,0.031,0.032,0.031;dbNSFP\_VindijiaNeandertal=C/C;dbNSFP\_bStatistic=620;dbNSFP\_bStatistic\_converted\_rankscore=0.66037;dbNSFP\_cds\_strand=-,-,-,-,-;dbNSFP\_clinvar\_MedGen\_id=C0023522\_CN169374\_CN517202;dbNSFP\_clinvar\_OMIM\_id=250100;dbNSFP\_clinvar\_Orphanet\_id=ORPHA512;dbNSFP\_clinvar\_clnsig=Benign;dbNSFP\_clinvar\_hgvs=NC\_000022.11:g.50625611G>C;dbNSFP\_clinvar\_id=21184;dbNSFP\_clinvar\_review=criteria\_provided,\_multiple\_submitters,\_no\_conflicts;dbNSFP\_clinvar\_trait=Metachromatic\_leukodystrophy\_not\_specified\_not\_provided;dbNSFP\_clinvar\_var\_source=HGMD:CM910052\_Illumina\_Clinical\_Services\_Laboratory,Illumina:31297;dbNSFP\_codon\_degeneracy=0,0,0,0,0;dbNSFP\_codonpos=2,2,2,2,2;dbNS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