# Download ANNOVAR

wget http://www.openbioinformatics.org/annovar/download/0wgxR2rIVP/annovar.latest.tar.gz

tar -zxvf annovar.latest.tar.gz

# Choose one

perl retrieve\_seq\_from\_fasta.pl -format genericGene -seqdir humandb/hg19\_seq/ humandb/hg19\_wgEncodeGencodeBasicV19.txt &

perl retrieve\_seq\_from\_fasta.pl -format genericGene -seqdir humandb/hg19\_seq/ -outfile humandb/hg19\_wgEncodeGencodeBasicV19Mrna.fa humandb/hg19\_wgEncodeGencodeBasicV19.txt &

## Download gene annotation database (for hg19 build) and save to humandb/ directory

# 1000g

perl annotate\_variation.pl -buildver hg19 -downdb -webfrom annovar 1000g2015aug humandb/ &

# dbSNP

perl annotate\_variation.pl -buildver hg19 -downdb -webfrom annovar snp138 humandb/ &

# dbNSFP (SIFT, PolyPhen2, and others)

perl annotate\_variation.pl -buildver hg19 -downdb -webfrom annovar dbnsfp33a humandb/ &

# download whole-genome FASTA files first before downloading GENCODE

perl annotate\_variation.pl -build hg19 -downdb seq humandb/hg19\_seq/ &

# GENCODE

perl annotate\_variation.pl -build hg19 -downdb wgEncodeGencodeBasicV19 humandb/ &

# refGene

perl annotate\_variation.pl -buildver hg19 -downdb -webfrom annovar refGene humandb/ &

# TFBS

perl annotate\_variation.pl -buildver hg19 -downdb tfbsConsSites humandb/ &

# main annotation code

perl table\_annovar.pl merged.6samples.vcf humandb/ -buildver hg19 -out 6samples -vcfinput -remove -protocol 1000g2015aug\_all,1000g2015aug\_eur,1000g2015aug\_amr,1000g2015aug\_afr,1000g2015aug\_eas,1000g2015aug\_sas,snp138,dbnsfp33a,wgEncodeGencodeBasicV19,refGene,tfbsConsSites -operation f,f,f,f,f,f,f,f,g,g,r -nastring . &>6samples\_log.txt &

perl table\_annovar.pl snp.vcf -buildver hg19 -out snp -vcfinput -remove –protocol gerp++gt2 -operation f -nastring . &>6samples\_log.txt &

perl table\_annovar.pl snp.vcf humandb/ -buildver hg19 -out gerp –vcfinput gerp++gt2 &

perl table\_annovar.pl snp.vcf -buildver hg19 -out gerp -vcfinput -remove -protocol gerp++gt2, -operation g, -nastring . humandb/ &>6samples\_log.txt &

perl table\_annovar.pl merged.6samples.vcf -buildver hg19 -out gerp -vcfinput -remove -protocol gerp++gt2, -operation g, -nastring . humandb/ &>6samples\_log.txt &

perl table\_annovar.pl snp.vcf -buildver hg19 -out cadd -vcfinput -remove -protocol cadd, -operation g, -nastring . humandb/ &>6samples\_log.txt &