CLCG ANNOTATION

CLCG Annotated File Header	Explanation
VARIATION	Genomic Coordinates for Variation
#OBSERVED	Number of aligned sequencing reads in which the variant was observed
#COVERED	Total depth of coverage for position at which the variant was observed
UI HET/HOMOZ	Heterozygous/Homozygous call by Univ of Iowa CLCG
GATK HET/HOMOZ	Heterozygous/Homozygous call by GATK pipeline
DEPTH	GATK depth of coverage for position at which the variant was observed
Q_VAR	GATK Quality of Variant Call -- Phred-like probability of variant being present
Q_GT	GATK quality of genotyping -- used primarily for when genotyping multiple individuals in one run
GENE	Gene name for variant
HGVS_NT	Variant in HGVS nucleotide format
HGVS PROTEIN	Variant in HGVS protein format
EXON/INTRON	Exon or intron where the variant is located
SYNONYMOUS	Indicates whether variant is synonymous, non-synonymous, indel, or splice site
dbSNP	RS# from the dbSNP database
Minor Allele Frequency	MAF from the dbSNP database
BLOSUM	BLOSUM pathogenicity score (<0 is more likely to be damaging)
POLYPHEN	Polyphen-formatted variant -- ready to be pasted in to online polyphen tool
SPLICE	Indicates if the variant is a splice variant
DIST_FROM_SPLICE	Distance the variant lies from a splice site
1KG_REF_ALLELE	Reference allele from the 1000 Genomes Project
1KG_REF_COUNT	Reference alelle count from the 1000 Genomes Project
1KG_ALT_ALLELE	Alternative allele from the 1000 Genomes Project
1KG_ALT_COUNT	Alternative alelle count from the 1000 Genomes Project
LUCAMP_HET_COUNT	Number of heterozygous carriers of this variant from the LuCamp et al dataset
LUCAMP_HOMOZ_ALT_COUNT	Number of homozygous carriers of this variant from the LuCamp et al dataset
LVD_NUM_SAMPLES	Number of samples in local variation database
LVD_NUM_ALLELES	Number of alleles in local variation database

ANNOVAR ANNOTATION

ANNOVAR Annotated File Header	Explanation
Func	Function of the variant -- exonic, intronic, UTR, etc.
Gene	Gene name for variant
ExonicFunc	If the variant is exonic, synonymous, non-synonymous, indel, etc.
AAChange	If exonic, variant change in nucleotide and protein format
Conserved	
SegDup	Indicates if the variant is located in a segmental duplication region
ESP5400_ALL	MAF in Exome Sequencing Project dataset (5,400 exomes) for all populations
1000g2012feb_ALL	MAF in 1000Genomes February 2012 release
dbSNP135	RS# from the dbSNP database
AVSIFT	SIFT Pathogenicity score: closer to 0 is more damaging
LJB_PhyloP	Pathogenicity score from dbNSFP: conserved > 0.95, not conserved < 0.95
LJB_PhyloP_Pred	Pathogenicity call from dbNSFP: C - conserved, N - not conserved
LJB_SIFT	Pathogenicity score from dbNSFP: tolerated < 0.95, deleterious > 0.95
LJB_SIFT_Pred	Pathogenicity call from dbNSFP: T - tolerated, D - deleterious
LJB_PolyPhen2	Pathogenicity score from dbNSFP: probably damaging > 0.85, possibly damaging 0.85-0.15, benign < 0.15
LJB_PolyPhen2_Pred	Pathogenicity call: D - probably damaging, P - possibly damaging, B - benign
LJB_LRT	Pathogenicity probability score from dbNSFP: closer to 1 is more likely to be damaging -- see below
LJB_LRT_Pred	Pathogenicity score from dbNSFP: D - deleterious fulfills the following: (i) from a codon defined by LRT as significantly constrained (LRTorio0.001 and oo1), (ii) from a site with Z10 eutherian mammals alignments, and (iii) the alternative AA is not presented in any of the eutherian mammals N - otherwise neutral
LJB_MutationTaster	Pathogenicity probability score from dbNSFP: closer to 1 is more likely to be damaging -- see below
LJB_MutationTaster_Pred	Pathogenicity score from dbNSFP: automatically calculated categories: ‘‘disease_causing_automatic,’’ ‘‘disease_causing,’’ ‘‘polymorphism,’’ and ‘‘polymorphism_automatic,’’ which we coded as ‘‘A,’’ ‘‘D,’’ ‘‘N,’’ and ‘‘P,’’
LJB_GERP++	Nucleotide conservation score from dbNSFP GERP: Higher number is more conserved, > 0 is generally conserved
Chr	Chromosome of variant
Start	Start coordinate of variant
End	End coordinate of variant
Ref	Reference allele for variant
Obs	Observed alllele for variant
Otherinfo	Other information