Name	Description
Symbol	Gene Symbol
Exonic Func	Feature location of the variant (exonic, splicing)
Gene Func	Functional consequences of the variant (nonsynonymous SNV, synonymous SNV, frameshift insertion, frameshift deletion, nonframeshift insertion, nonframeshift deletion, frameshift block substitution, nonframshift block substitution)
Aachange	Amino acid change
chrom	Chromosome name
pos	Chromosomal position
ref	Reference allele
alt	Alternative observed alleles
type	SNV or INDEL
qual	Global score of the calling
filter	Result of the global quality filter after the gaussian mixture model: 'PASS'=passed
vqslod	Log odds ratio of being a true variant versus being false under the trained gaussian mixture model
Variation Class	Classification of the variation (I,II,IIIIV,V)
<sample name> ZYG	Decoded genotype
<sample name> GT	Called genotype
<sample name> N REF reads	N reads supporting the reference allele
<sample name> N ALT reads	N reads supporting the alternative allele
<sample name> N tot reads	Total number of reads at position
<sample name> Perc ALT reads	Percentage of reads supporting the alternative allele
<sample name> GQ	Genotype Quality
Avsift	SIFT pre-calculated P-value: if < 0.05 the mutation is considered deleterious
LJB SIFT	SIFT from dbNSF (between 0 and 1, the higher the more deleterious)
LJB PolyPhen2	PolyPhen2 from dbNSF (between 0 and 1, the higher the more deleterious)
LJB LRT	LRT from dbNSF (between 0 and 1, the higher the more deleterious)
LJB Mutation Taster	MutationTaster from dbNSF (between 0 and 1, the higher the more deleterious)
LJB PhyloP	PhyloP conservation score
LJB PhyloP Pred	PlyloP prediction: C (conserved) if score > 0.95, otherwise N (non conserved)
LJB Gerp++	Gerp++ conservation score (the higher the more conserved)
Conserved	Conservation score from UCSC 46 species alignment: range 0-1000, the higher the more conserved
freq ESP6500	Frequency in NCBI 6500 exomes project
freq1000g 2012apr ALL	Frequency in 1000 genomes data (release apr 2012), all populations together
freq1000g 2012apr AFR	Frequency in 1000 genomes data (release apr 2012), subpop
freq1000g 2012apr AMR	Frequency in 1000 genomes data (release apr 2012), subpop
freq1000g 2012apr ASN	Frequency in 1000 genomes data (release apr 2012), subpop
freq1000g 2012apr EUR	Frequency in 1000 genomes data (release apr 2012), subpop
dbsnp137	dbSNP 137 ID
dbSNP137 NonFlagged	dbSNP 137 after removing those flagged SNPs (SNPs < 1% minor allele frequency (MAF) (or unknown), mapping only once to reference assembly, flagged in dbSNP as "clinically associated")
dbSNP137 Observed Allele	dbSNP 137 Observed Allele
OMIM	OMIM ID
freq OMIM:X	Allele frequency in patients affected by disease OMIM:X
freq OMIM:X Controls	Allele frequency in unaffected controls of disease OMIM:X
freq OMIM 1	Allele frequency in patients affected by disease OMIM ID1
freq OMIM ..	Allele frequency in patients affected by disease OMIM ID...
freq OMIM:N	Allele frequency in patients affected by disease OMIM:N