Variants

reference_name : id from reference genome : STRING
start : starting position : INTEGER
end : ending position: INTEGER
reference_base: reference from genome: STRING
alternate_base: base found in patient: STRING
quality: Phred score: FLOAT
filter: PASS if consition is satisfied :INTEGER
names: unique ID for variants: STRING
call: per sampled measurement : RECORD
call.call_set_id: The id of the callset : STRING
call.call_set_name: Sample identifier: STRING
call.genotype: List of genotypes: INTEGER
call.phaseset: If this value is null, the data is unphased. Otherwise it is phased: STRING
call.genotype_likelyhood: List of genotype likelyhood: FLOAT
call.DP: High quality bases: INTEGER
call.DS: Genotype dosage from MaCH/Thunder: FLOAT
call.FT:STRING
call.GQ: Genotype quality: STRING
call.PL: List of Phred-scaled genotype likelihoods, number of values is (#ALT+1)*(#ALT+2)/2: INTEGER
call.sp:Phred-scaled strand bias P-value:INTEGER
AA: Ancestral allele: STRING
AC: Alternate allele count: INTEGER
AC1:<ID=AC1,Number=1,Type=Float,Description="Max-likelihood estimate of the first ALT allele count (no HWE assumption):INTEGER
AF:Global Allele Frequency based on AC/AN:FLOAT
AF1:Max-likelihood estimate of the first ALT allele frequency (assuming HWE):FLOAT
AFR_AF:Allele Frequency for samples from AFR based on AC/AN:FLOAT
AMR_AF:Allele Frequency for samples from AMR based on AC/AN:FLOAT
AN:Total allele count: INTEGER
ASN_AF:Allele Frequency for samples from ASN based on AC/AN:FLOAT
AVGPOST:Average posterior probability from MaCH/Thunder:FLOAT
CIEND:Confidence interval around END for imprecise variants:INTEGER
CPOST:Confidence interval around POS for imprecise variants:INTEGER
DP: Raw read depth: INTEGER
DP4:high-quality ref-forward bases, ref-reverse, alt-forward and alt-reverse bases:INTEGER
erate:Per-marker Mutation rate from MaCH/Thunder:FLOAT
eur_af:Allele Frequency for samples from EUR based on AC/AN:FLOAT
fq:Phred probability of all samples being the same:FLOAT
G3:ML estimate of genotype frequencies:FLOAT
homlen:Length of base pair identical micro-homology at event breakpoints:INTEGER
homseq:Sequence of base pair identical micro-homology at event breakpoints:STRING
HWE:Chi^2 based HWE test P-value based on G3:FLOAT
LDAF:MLE Allele Frequency Accounting for LD:FLOAT
MQ:Root-mean-square mapping quality of covering reads:INTEGER
PV4:P-values for strand bias, baseQ bias, mapQ bias and tail distance bias:FLOAT
RSQ:Genotype imputation quality from MaCH/Thunder:FLOAT
SNPSOURCE:indicates if a snp was called when analysing the low coverage or exome alignment data:STRING
SOURCE:STRING
SVLEN:Difference in length between REF and ALT alleles:INTEGER
SVTYPE:Type of structural variant:STRING
theta:Per-marker Transition rate from MaCH/Thunder:FLOAT
VT:indicates what type of variant the line represents:STRING

Sample_info

sample: Sample_id : STRING : PRIMARY KEY
family_id: Family_ID: STRING
population: 3 letter population code: STRING
population_description: description of the population: STRING
Gender: gender: STRING
relationship: relationship to other members: STRING
Unexpected_Parent_Child: sample id for unexpected parent child relationships: STRING
Non_Paternity:sample ids for annotated non paternal relationships: STRING
Siblings:sample ids for any siblings: STRING
Grandparents:sample ids for any grand parents: STRING
Avuncular:sample ids for any avuncular relationships:STRING
Half_Siblings:sample ids for any half siblings:STRING
Unknown_Second_Order:sample ids for any unknown second order relations:STRING
Third_Order:sample ids for any third order cryptic relations:STRING
In_Low_Coverage_Pilot:The sample is in the low coverage pilot experiment:BOOLEAN
LC_Pilot_Platforms:low coverage pilot sequencing platforms:STRING
LC_Pilot_Centers:low coverage pilot sequencing centers:STRING
In_High_Coverage_Pilot:The sample is in the high coverage pilot:BOOLEAN
HC_Pilot_Platforms:high coverage sequencing platforms:STRING
HC_Pilot_Centers:high coverage sequencing centers:STRING
In_Exon_Targetted_Pilot:The Sample is in the exon targetted pilot experiment:BOOLEAN
ET_Pilot_Platforms:exon targetted sequencing platforms:STRING
ET_Pilot_Centers:exon targetted sequencing centers:STRING
Has_Sequence_in_Phase1:BOOLEAN
Phase1_LC_Platform:phase1 low coverage sequencing platforms:STRING
Phase1_LC_Centers:phase1 low coverage sequencing centers:STRING
Phase1_E_Platform:phase1 exome sequencing platforms:STRING
Phase1_E_Centers:phase1 exome sequencing centers:STRING
In_Phase1_Integrated_Variant_Set:BOOLEAN
Has_Phase1_chrY_SNP:BOOLEAN
Has_phase1_chrY_Deletions:BOOLEAN
Has_phase1_chrMT_SNPs:BOOLEAN
Main_project_LC_Centers:STRING
Main_project_LC_platform:STRING
Total_LC_Sequence:FLOAT
LC_Non_Duplicated_Aligned_Coverage:FLOAT
Main_Project_E_Centers:STRING
Main_Project_E_Platform:STRING
Total_Exome_Sequence:FLOAT
X_Targets_Covered_to_20x_or_greater:FLOAT
VerifyBam_E_Omni_Free:FLOAT
VerifyBam_E_Affy_Free:FLOAT
VerifyBam_E_Omni_Chip:FLOAT
VerifyBam_E_Affy_Chip:FLOAT
VerifyBam_LC_Omni_Free:FLOAT
VerifyBam_LC_Affy_Free:FLOAT
VerifyBam_LC_Omni_Chip:FLOAT
VerifyBam_LC_Affy_Chip:FLOAT
LC_Indel_Ratio: If the ratio is higher than 5 the sample is withdrawn:FLOAT
E_Indel_Ratio:If ratio > 5 is withdrawn:FLOAT
LC_Passed_QC:BOOLEAN
E_Passed_QC:BOOLEAN
In_Final_Phase_Variant_Calling:BOOLEAN
Has_Omni_Genotypes:BOOLEAN
Has_Axiom_Genotypes:BOOLEAN
Has_Affy_6_0_Genotypes:BOOLEAN
Has_Exome_LOF_Genotypes:BOOLEAN
EBV_Coverage:FLOAT
DNA_Source_from_Coriell:STRING
Has_Sequence_from_Blood_in_Index:BOOLEAN
Super_Population:STRING
Super_Population_Description:STRING

Pedigree

family_id:STRING
individual_ID:STRING: PRIMARY_KEY
paternal_ID:STRING
maternal_ID:STRING
gender:INTEGER
phenotype:INTEGER
population:STRING
relationship:STRING
siblings:STRING
second_order:STRING
thirs_order:STRING
other_comments:STRING