Variant Annotation Tool Update

LH

2/8/11

1000 low coverage data

- SNPs / Indels (six files)
  - CEU
  - CHBJPT
  - YRI
- VCF files include genotype information
- Annotation performed using snpMapper and indelMapper
- Merged resulting VCF files

VCF

```
##ANNOTATION=gencode.interval
#CHROM  POS     ID      REF     ALT     QUAL    FILTER  INFO    ANNOTATION      FORMAT  SAMPLE
chrM    3548    .       A       G       .       PASS    .       1:MT-ND1:ENSG00000198888:+:nonsynonymous:1/1:MT-ND1-201:ENST00000361390:954_241_81_I->V GT      1/1
chrM    6024    .       G       A       .       PASS    .       1:MT-CO1:ENSG00000198804:+:synonymous:1/1:MT-CO1-201:ENST00000361624:1539_120_40_E->E   GT      0/1
chrM    8015    .       A       T       .       PASS    .       1:MT-CO2:ENSG00000198712:+:synonymous:1/1:MT-CO2-201:ENST00000361739:681_429_143_V->V   GT      1/1
chrM    8252    .       G       A       .       PASS    .       1:MT-CO2:ENSG00000198712:+:synonymous:1/1:MT-CO2-201:ENST00000361739:681_666_222_G->G   GT      1/1
chrM    8702    .       G       A       .       PASS    .       1:AF347015.1:ENSG00000198899:+:nonsynonymous:1/1:AF347015.1-201:ENST00000361899:678_175_59_A->T GT      1/1
chrM    9378    .       G       A       .       PASS    .       1:MT-CO3:ENSG00000198938:+:prematureStop:1/1:MT-CO3-201:ENST00000362079:781_171_57_W->* GT      1/1
chrM    9541    .       C       T       .       PASS    .       1:MT-CO3:ENSG00000198938:+:synonymous:1/1:MT-CO3-201:ENST00000362079:781_334_112_L->L   GT      1/1
chrM    10399   .       G       A       .       PASS    .       1:MT-ND3:ENSG00000198840:+:nonsynonymous:1/1:MT-ND3-201:ENST00000361227:346_340_114_A->T        GT      1/1
chrM    10820   .       G       A       .       PASS    .       1:MT-ND4:ENSG00000198886:+:synonymous:1/1:MT-ND4-201:ENST00000361381:1378_60_20_K->K    GT      1/1
chrM    10874   .       C       T       .       PASS    .       1:MT-ND4:ENSG00000198886:+:synonymous:1/1:MT-ND4-201:ENST00000361381:1378_114_38_P->P   GT      1/1
chrM    11018   .       C       T       .       PASS    .       1:MT-ND4:ENSG00000198886:+:synonymous:1/1:MT-ND4-201:ENST00000361381:1378_258_86_S->S   GT      1/1
chrM    11720   .       A       G       .       PASS    .       1:MT-ND4:ENSG00000198886:+:synonymous:1/1:MT-ND4-201:ENST00000361381:1378_960_320_G->G  GT      1/1
chrM    11723   .       C       T       .       PASS    .       1:MT-ND4:ENSG00000198886:+:synonymous:1/1:MT-ND4-201:ENST00000361381:1378_963_321_L->L  GT      1/1
chrM    12697   .       T       C       .       PASS    .       1:MT-ND5:ENSG00000198786:+:synonymous:1/1:MT-ND5-201:ENST00000361567:1809_360_120_Y->Y  GT      1/1
chrM    12706   .       T       C       .       PASS    .       1:MT-ND5:ENSG00000198786:+:synonymous:1/1:MT-ND5-201:ENST00000361567:1809_369_123_I->I  GT      1/1
chrM    12851   .       G       A       .       PASS    .       1:MT-ND5:ENSG00000198786:+:nonsynonymous:1/1:MT-ND5-201:ENST00000361567:1809_514_172_V->I       GT      1/1
chrM    14213   .       C       T       .       PASS    .       1:MT-ND6:ENSG00000198695:-:synonymous:1/1:MT-ND6-201:ENST00000361681:522_462_154_V->V   GT      1/1
chrM    14581   .       G       A       .       PASS    .       1:MT-ND6:ENSG00000198695:-:synonymous:1/1:MT-ND6-201:ENST00000361681:522_94_32_L->L     GT      1/1
chrM    14767   .       T       C       .       PASS    .       1:MT-CYB:ENSG00000198727:+:nonsynonymous:1/1:MT-CYB-201:ENST00000361789:1135_20_7_I->T  GT      1/1
chrM    14906   .       A       G       .       PASS    .       1:MT-CYB:ENSG00000198727:+:nonsynonymous:1/1:MT-CYB-201:ENST00000361789:1135_159_53_I->M        GT      1/1
chrM    15219   .       A       G       .       PASS    .       1:MT-CYB:ENSG00000198727:+:nonsynonymous:1/1:MT-CYB-201:ENST00000361789:1135_472_158_T->A       GT      1/1
chrM    15302   .       A       G       .       PASS    .       1:MT-CYB:ENSG00000198727:+:synonymous:1/1:MT-CYB-201:ENST00000361789:1135_555_185_L->L  GT      1/1
```

VCF annotation field:

alleleNumber:geneName:geneId:strand:type:fraction:transcripts

Results: 1000genomes_lowCoverage

Show 25 entries

Search:

Gene ID	Gene name	Number of transcripts	Number of synonymous SNPs	Number of nonsynonymous SNPs	Number of prematureStop SNPs	Number of removedStop SNPs	Number of spliceOverlaps	Number of insertions	Number of deletions	Details
ENSG00000004897	CDC27	3	4	3	0	0	10	0	0	Link
ENSG00000205592	MUC19	8	28	65	4	1	7	1	3	Link
ENSG00000234603	AC013460.1	3	11	14	0	0	5	1	3	Link
ENSG00000100336	APOL4	12	6	17	1	0	4	1	0	Link
ENSG00000178852	C17orf57	2	5	6	4	0	4	0	0	Link
ENSG00000197558	SSPO	10	52	105	5	0	4	5	4	Link
ENSG00000204520	MICA	10	7	27	0	0	4	1	2	Link
ENSG00000232769	AC137600.1	2	2	24	0	0	4	3	5	Link
ENSG00000233389	AC011543.1	3	13	21	1	0	4	3	7	Link
ENSG00000148408	CACNA1B	11	16	9	0	0	3	0	1	Link
ENSG00000179344	HLA-DQB1	6	31	49	0	0	3	0	0	Link
ENSG00000189157	FAM47E	1	0	6	0	0	3	0	0	Link
ENSG00000202582	AC131157.4	2	12	32	0	0	3	4	5	Link
ENSG00000204296	C6orf10	7	6	21	1	0	3	0	2	Link
ENSG00000205783	AC091435.2	3	6	19	0	0	3	2	7	Link
ENSG00000205785	AC091435.1	6	12	24	1	0	3	4	7	Link
ENSG00000231499	AF186191.1	1	2	11	0	0	3	0	4	Link
ENSG00000232190	AC005756.1	1	6	14	0	0	3	3	3	Link
ENSG00000236908	AC019043.1	1	5	9	0	0	3	2	2	Link
ENSG00000005206	AC004410.1	4	13	2	0	0	2	1	0	Link
ENSG00000010361	FUZ	4	3	5	0	0	2	0	1	Link
ENSG00000059588	TARBP1	1	12	12	0	0	2	0	0	Link
ENSG00000076944	STXBP2	4	16	13	2	0	2	3	1	Link
ENSG00000078674	PCM1	5	9	13	0	0	2	0	0	Link
ENSG00000083635	NUFIP1	1	2	2	0	0	2	0	0	Link

Showing 1 to 25 of 18,382 entries

First Previous 1 2 3 4 5 Next Last

1000genomes_lowCoverage: gene summary for FUZ [ENSG00000010361]

External links:

[UCSC genome browser] [Ensembl genome browser] [Gene Cards]

Transcript summary

Transcript name	Transcript ID	Chromosome	Strand	Start	End	Number of exons	Transcript length
FUZ-203	ENST00000421740	chr19	-	55006214	55008175	6	705
FUZ-204	ENST00000445575	chr19	-	55002152	55008175	13	1161
FUZ-202	ENST00000377092	chr19	-	55002222	55008175	10	1146
FUZ-201	ENST00000313777	chr19	-	55002222	55008175	11	1254

Graphical representation of genetic variants

LEGEND FOR VARIATION TYPES:

spliceOverlap synonymous nonsynonymous prematureStop removedStop insertion deletion substitution

Detailed summary of variants

Chromosome	Position	Reference allele	Alternate allele	Identifier	Type	Fraction of transcripts affected	Transcripts	Transcript details	Alternate allele frequencies			Genotypes
									CEU	CHBJPT	YRI	
chr19	55006228	AAGAG	A	.	deletion	1/4	ENST00000421740	705_692	0.025	0.000	0.000	Link
chr19	55002239	G	T	.	nonsynonymous	2/4	ENST00000313777 ENST00000377092	1254_1238_413_A->D 1146_1130_377_A->D	0.000	0.000	0.051	Link
chr19	55002278	G	A	rs12610577	nonsynonymous	2/4	ENST00000313777 ENST00000377092	1254_1199_400_T->I 1146_1091_364_T->I	0.000	0.117	0.085	Link
chr19	55003512	G	T	.	nonsynonymous	3/4	ENST00000445575 ENST00000313777 ENST00000377092	1161_1004_335_T->N 1254_1004_335_T->N 1146_896_299_T->N	0.008	0.000	0.000	Link
chr19	55003860	G	A	rs11557714	synonymous	3/4	ENST00000445575 ENST00000313777 ENST00000377092	1161_819_273_D->D 1254_819_273_D->D 1146_711_237_D->D	0.033	0.000	0.000	Link
chr19	55004465	C	T	rs2305921	synonymous	3/4	ENST00000445575 ENST00000313777 ENST00000377092	1161_672_224_L->L 1254_672_224_L->L 1146_564_188_L->L	0.125	0.133	0.136	Link
chr19	55006232	G	A	.	nonsynonymous	1/4	ENST00000421740	705_688_230_L->F	0.008	0.000	0.000	Link
chr19	55006282	A	G	.	spliceOverlap	1/4	ENST00000421740	705	0.050	0.000	0.000	Link
chr19	55006283	G	A	.	spliceOverlap	1/4	ENST00000421740	705	0.100	0.058	0.000	Link
chr19	55006519	G	A	rs35499921	synonymous	4/4	ENST00000445575 ENST00000313777 ENST00000377092 ENST00000421740	1161_405_135_I->I 1254_405_135_I->I 1146_297_99_I->I 705_405_135_I->I	0.000	0.000	0.025	Link
chr19	55008076	C	A	rs35138412	nonsynonymous	4/4	ENST00000445575 ENST00000313777 ENST00000377092 ENST00000421740	1161_100_34_A->S 1254_100_34_A->S 1146_100_34_A->S 705_100_34_A->S	0.000	0.000	0.051	Link

Variant summary

Chromosome	Position	Reference allele	Alternate allele
chr19	55004465	C	T

Genotype information

CEU	CHBJPT	YRI
RefCount = 105, AltCount = 15	RefCount = 104, AltCount = 16	RefCount = 102, AltCount = 16
NA06985: 0|0	NA18526: 0|0	NA18486: 0|0
NA06986: 0|0	NA18532: 0|0	NA18489: 0|0
NA06994: 0|0	NA18537: 0|0	NA18498: 0|0
NA07000: 0|0	NA18542: 0|1	NA18499: 0|1
NA07037: 0|0	NA18545: 0|0	NA18501: 0|0
NA07051: 1|0	NA18547: 0|0	NA18502: 0|1
NA07346: 0|0	NA18550: 0|0	NA18504: 0|0
NA07347: 0|0	NA18552: 0|1	NA18505: 0|0
NA07357: 0|0	NA18555: 0|0	NA18507: 0|1
NA10847: 0|0	NA18558: 0|0	NA18508: 0|0
NA10851: 0|0	NA18561: 0|0	NA18510: 0|0
NA11829: 0|0	NA18562: 0|1	NA18511: 0|0
NA11830: 0|0	NA18563: 0|0	NA18516: 0|0
NA11831: 0|0	NA18564: 0|0	NA18517: 0|0
NA11832: 0|0	NA18566: 0|0	NA18519: 0|1
NA11840: 0|0	NA18570: 0|1	NA18520: 1|0
NA11881: 0|1	NA18571: 0|1	NA18522: 0|0
NA11894: 0|0	NA18572: 0|0	NA18523: 0|0
NA11918: 0|0	NA18573: 0|0	NA18853: 0|1
NA11919: 1|0	NA18576: 0|0	NA18856: 0|0
NA11920: 0|0	NA18577: 0|0	NA18858: 0|1
NA11931: 0|0	NA18579: 1|0	NA18861: 0|0
NA11992: 1|0	NA18582: 0|0	NA18870: 0|1
NA11993: 0|1	NA18592: 0|0	NA18871: 0|0
NA11994: 0|1	NA18593: 0|0	NA18907: 0|0
NA11995: 0|0	NA18603: 0|1	NA18909: 0|0
NA12003: 0|0	NA18605: 1|0	NA18912: 0|0
NA12004: 0|0	NA18608: 0|0	NA18916: 1|1
NA12005: 0|0	NA18609: 0|0	NA19093: 0|0
NA12006: 0|1	NA18638: 0|1	NA19098: 0|0
NA12043: 0|0	NA18940: 0|1	NA19099: 0|0
NA12044: 0|0	NA18942: 0|0	NA19102: 0|1
NA12045: 1|0	NA18943: 0|0	NA19108: 0|0
NA12144: 0|0	NA18944: 0|0	NA19114: 1|0
NA12154: 0|0	NA18945: 0|0	NA19116: 0|1
NA12155: 0|0	NA18947: 0|0	NA19119: 0|0
NA12156: 0|0	NA18948: 0|0	NA19129: 1|0
NA12234: 0|0	NA18949: 0|0	NA19131: 0|0
NA12249: 0|1	NA18951: 0|0	NA19137: 0|0
NA12287: 0|0	NA18952: 0|0	NA19138: 0|0
NA12414: 0|0	NA18953: 0|0	NA19141: 0|0
NA12489: 0|0	NA18956: 1|0	NA19143: 1|0
NA12716: 0|1	NA18959: 0|0	NA19144: 0|0
NA12717: 0|0	NA18960: 0|0	NA19147: 0|0
NA12749: 0|1	NA18961: 1|0	NA19152: 0|0
NA12750: 0|0	NA18964: 0|0	NA19153: 0|0
NA12751: 0|0	NA18965: 0|0	NA19159: 0|0
NA12760: 0|0	NA18967: 1|0	NA19160: 1|0
NA12761: 0|0	NA18968: 0|0	NA19171: 0|0
NA12762: 0|0	NA18969: 0|1	NA19172: 0|0
NA12763: 1|0	NA18970: 0|0	NA19190: 0|0
NA12776: 0|0	NA18971: 0|0	NA19200: 0|0
NA12812: 0|0	NA18972: 1|0	NA19201: 0|0
NA12813: 1|1	NA18973: 1|0	NA19204: 0|0
NA12814: 0|0	NA18974: 0|0	NA19207: 0|0
NA12815: 0|0	NA18975: 0|0	NA19209: 0|0
NA12828: 0|0	NA18976: 0|0	NA19210: 0|0
NA12872: 0|0	NA18980: 0|0	NA19225: 0|0
NA12873: 1|0	NA18981: 0|0	NA19257: 0|0
NA12874: 0|0	NA19005: 0|0	