



Supplementary Figure 2

A.

artem@glitch[~/Desktop]	
43 ##INFO=<ID=GNO,Number=0,Type=Flag,Description="Genotypes available. The variant has individual genotype (in SubInd table).">	
44 ##INFO=<ID=KGPhase1,Number=0,Type=Flag,Description="1000 Genome phase 1 (incl. June Interim phase 1)">	
45 ##INFO=<ID=KGPhase3,Number=0,Type=Flag,Description="1000 Genome phase 3">	
46 ##INFO=<ID=CDA,Number=0,Type=Flag,Description="Variation is interrogated in a clinical diagnostic assay">	
47 ##INFO=<ID=LSD,Number=0,Type=Flag,Description="Submitted from a locus-specific database">	
48 ##INFO=<ID=MTF,Number=0,Type=Flag,Description="Microattribution/third-party annotation(TPA:GWAS,PAGE)">	
49 ##INFO=<ID=OM,Number=0,Type=Flag,Description="Has OMIM/OMIA">	
50 ##INFO=<ID=NOC,Number=0,Type=Flag,Description="Contig allele not present in variant allele list. The reference sequence allele at the mapped positi	
51 ##INFO=<ID=NTD,Number=0,Type=Flag,Description="Is Withdrawn by submitter. If one member ss is withdrawn by submitter, then this bit is set. If all	
52 ##INFO=<ID=NOV,Number=0,Type=Flag,Description="Rs cluster has non-overlapping allele sets. True when rs set has more than 2 alleles from different	
53 ##FILTER=<ID=NC,Description="Inconsistent Genotype Submission For At Least One Sample">	
54 ##INFO=<ID=CAF,Number=1,Type=String,Description="An ordered, comma delimited list of allele frequencies based on 1000Genomes, starting with the ref	
55 ##INFO=<ID=COMMON,Number=1,Type=Integer,Description="RS is a common SNP. A common SNP is one that has at least one 1000Genomes population with a n	
56 ##INFO=<ID=TOPMED,Number=0,Type=String,Description="An ordered, comma delimited list of allele frequencies based on TOPMED, starting with the refer	
#CHROM	POS ID REF ALT QUAL FILTER INFO
1	10177 rs367896724 A G RS=367896724;RSPPOS=10177;dbSNPBuildID=138;SSR=0;SAO=0;VP=0x050000020005170026000200
1	10352 rs555500075 A G RS=555500075;RSPPOS=10352;dbSNPBuildID=142;SSR=0;SAO=0;VP=0x050000020005170026000200
1	10352 rs145072688 A G RS=145072688;RSPPOS=10353;dbSNPBuildID=134;SSR=0;SAO=0;VP=0x050000020005000002000200
1	10616 rs376342519 C G RS=376342519;RSPPOS=10617;dbSNPBuildID=142;SSR=0;SAO=0;VP=0x05000002
1	10642 rs558604819 C G RS=558604819;RSPPOS=10642;dbSNPBuildID=142;SSR=0;SAO=0;VP=0x050000020005040024000100
1	11008 rs575272151 A G RS=575272151;RSPPOS=11008;dbSNPBuildID=142;SSR=0;SAO=0;VP=0x050000020005150024000100
1	11012 rs544419019 G C RS=544419019;RSPPOS=11012;dbSNPBuildID=142;SSR=0;SAO=0;VP=0x050000020005150024000100
1	11063 rs561109771 G C RS=561109771;RSPPOS=11063;dbSNPBuildID=142;SSR=0;SAO=0;VP=0x050000020005040024000100
1	13110 rs540538026 G C RS=540538026;RSPPOS=13110;dbSNPBuildID=142;SSR=0;SAO=0;VP=0x050000000005150026000100
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1	13118 rs62028691 G C RS=62028691;RSPPOS=13118;RV;dbSNPBuildID=130;SSR=0;SAO=0;VP=0x0500000000051401260001
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1	13289 rs538791886 G C RS=538791886;RSPPOS=13290;dbSNPBuildID=142;SSR=0;SAO=0;VP=0x050000000005040026000200
1	13380 rs571093408 G C RS=571093408;RSPPOS=13380;dbSNPBuildID=142;SSR=0;SAO=0;VP=0x050000000005040026000100
1	13445 rs558318514 G C RS=558318514;RSPPOS=13445;dbSNPBuildID=142;SSR=0;SAO=0;VP=0x050000000005040026000100
1	13453 rs568927457 G C RS=568927457;RSPPOS=13453;dbSNPBuildID=142;SSR=0;SAO=0;VP=0x050000000005040026000100
1	13483 rs554760071 G C RS=554760071;RSPPOS=13483;dbSNPBuildID=142;SSR=0;SAO=0;VP=0x050000000005040026000100
1	13550 rs554008981 G C RS=554008981;RSPPOS=13550;dbSNPBuildID=142;SSR=0;SAO=0;VP=0x050000000005040026000100

B.

artem@glitch[~/Desktop]	
1623 chr1 HAVANA transcript 1255178 1273833	gene_id "ENSG00000160007.20"; transcript_id "ENS
1624 chr1 HAVANA exon 1273666 1273833	gene_id "ENSG00000160007.20"; transcript_id "ENS
1625 chr1 HAVANA exon 1267862 1267992	gene_id "ENSG00000160007.20"; transcript_id "ENS
1626 chr1 HAVANA 5'UTR 1267862 1267992	gene_id "ENSG00000160007.20"; transcript_id "ENS
1627 chr1 HAVANA start_codon 1267990 1267992	gene_id "ENSG00000160007.20"; transcript_id "ENS
1628 chr1 HAVANA exon 1264783 1264830	gene_id "ENSG00000160007.20"; transcript_id "ENS
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1630 chr1 HAVANA exon 1263346 1263386	gene_id "ENSG00000160007.20"; transcript_id "ENS
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1632 chr1 HAVANA exon 1257208 1257310	gene_id "ENSG00000160007.20"; transcript_id "ENS
1633 chr1 HAVANA 5'UTR 1257208 1257310	gene_id "ENSG00000160007.20"; transcript_id "ENS
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1635 chr1 HAVANA 5'UTR 1256992 1257130	gene_id "ENSG00000160007.20"; transcript_id "ENS
1636 chr1 HAVANA exon 1256045 1256125	gene_id "ENSG00000160007.20"; transcript_id "ENS
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1642 chr1 HAVANA UTR 1255178 1255205	gene_id "ENSG00000160007.20"; transcript_id "ENS
1643 chr1 HAVANA gene 1275223 1280420	gene_id "ENSG00000230415.1"; gene_type "lincRNA"
1644 chr1 HAVANA transcript 1275223 1280420	gene_id "ENSG00000230415.1"; transcript_id "ENST
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1646 chr1 HAVANA exon 1279068 1279148	gene_id "ENSG00000230415.1"; transcript_id "ENST
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1650 chr1 HAVANA exon 1279068 1279393	gene_id "ENSG00000230415.1"; transcript_id "ENST
1651 chr1 HAVANA gene 1280436 1292019	gene_id "ENSG00000162572.20"; gene_type "protein"
1652 chr1 HAVANA transcript 1280436 1292019	gene_id "ENSG00000162572.20"; transcript_id "ENS
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1654 chr1 HAVANA 5'UTR 1280662 1280666	gene_id "ENSG00000162572.20"; transcript_id "ENS
1655 chr1 HAVANA start_codon 1280662 1280664	gene_id "ENSG00000162572.20"; transcript_id "ENS
1656 chr1 HAVANA exon 1281226 1281297	gene_id "ENSG00000162572.20"; transcript_id "ENS

C.

artem@glitch[~/Desktop]	
1 @HD	VN:1.0 SO:coordinate
2 @SQ	SN:chr13 LN:1023550
3 @RG	ID:MARTO LB:PLUMBERS SM:HG008BIT PL:ILLUMINA PU:SNES-ST64
4 @PG	ID:bowtie2 PN:bowtie2 VN:2.2.6 CL:"usr/bin/bowtie2-align-n -8bit -1 inpipe1 -2 inpipe2"
5	SNES1993.19247142 163 chr13 10001 10044 168
6	SNES1993.35201119 73 chr13 10001 10001 0
7	SNES1993.11914589 69 chr13 10001 10001 0
8	SNES1993.31258687 133 chr13 10001 95M61 0
9	SNES1993.32192276 133 chr13 10001 10001 0
10	SNES1993.35201119 133 chr13 10001 10001 0
11	SNES1993.35553638 69 chr13 10001 10001 0
12	SNES1993.55013408 69 chr13 10001 10001 0
13	SNES1993.11914589 153 chr13 10001 8D93M 0
14	SNES1993.31258687 89 chr13 10001 10001 0
15	SNES1993.32192276 89 chr13 10001 67534M 0
16	SNES1993.35553638 153 chr13 10001 10001 0
17	SNES1993.55013408 153 chr13 10001 10001 0
18	SNES1993.7932405 69 chr13 10003 10003 0
19	SNES1993.7932405 1177 chr13 10003 10001 0
20	SNES1993.19247142 83 chr13 10044 50M51S 10001 -168
21	SNES1993.49659117 97 chr13 10140 101M 10664 625
22	SNES1993.58751204 99 chr13 10143 101M 10352 234
23	SNES1993.36089268 99 chr13 10149 101M 10240 192
24	SNES1993.57860057 73 chr13 10149 101M 10149 0
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26	SNES1993.2803200 73 chr13 10151 100M1S 10151 0
27	SNES1993.2803200 537 chr13 10151 101M 10151 0
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30	SNES1993.56664346 137 chr13 10152 101M 10152 0
31	SNES1993.9345310 69 chr13 10152 101M 10152 0
32	SNES1993.9927629 133 chr13 10152 101M 10152 0
33	SNES1993.56664346 69 chr13 10152 101M 10152 0
34	SNES1993.26275897 99 chr13 10153 101M 10238 186

Supplementary Figure 3

