

Thousands of human mutation clusters are explained by short-range template switching

Ari Löytynoja

Data S2: TSM candidates discovered in NA12878 with short-read data.

The locus is given on the header line; different loci are separated by dashes. Top: template switch solution, template switch alignment and forward alignment. Middle: variants and genotypes for NA12878 in the HaplotypSV data and in the 1000 Genomes data; the latter also includes allele frequencies globally and among Europeans. Bottom: PacBio alignment of NA12878 reads.

Platinum & Chromium chr1:68086419-68086450

Switch process:

F1: L ACGGACTAACTAAGGAAGTTATCTTATTTATCTAAGAGCACAAGA 1
F3: 4 ACTTCTTGAGGCCAAAGATAATTTCTATGTTACTTGAAACTAG R
RF: ACGGACTAACTAAGGAAGTTATCTTATTTATCTAAGAGCACAAGAGATAGAGCATTGTGGTGAAGAGTTTCTATCACTTCTTGAGGCCAAAGATAATTTCTATGTTACTTGAAACTAG
RR: TGCCTGATTGATTCCTTCAATAGATAAAATAGATTCTCGTGTCTCTATCTCGTAACACCACTTCTCAAAGATAGTGAAGAACTCCGGTTTCTATTAAAGATACAATGAACCTTTGATC
F2: 3 CTATCTCGTAACACCACTTCTCAAAGATAG 2

Template-switch alignment:

REF ACGGACTAACTAAGGAAGTTATCTTATTTATCTAAGAGCACAAGA|GATAGAACTCTTCACCACAATGCTCTATC|ACTTCTTGAGGCCAAAGATAATTTCTATGTTACTTGAAACTAG
ALT ACGGACTAACTAAGGAAGTTATCTTATTTATCTAAGAGCACAAGA|GATAGAACTCTTCACCACAATGCTCTATC|ACTTCTTGAGGCCAAAGATAATTTCTATGTTACTTGAAACTAG

Forward alignment:

REF ACGGACTAACTAAGGAAGTTATCTTATTTATCTAAGAGCACAAGAGATAGAAacTctTcaccAcaAtgcTCTATCACTTCTTGAGGCCAAAGATAATTTCTATGTTACTTGAAACTAG
ALT ACGGACTAACTAAGGAAGTTATCTTATTTATCTAAGAGCACAAGAGATAGAGCATTGTGGTGAAGAGTTTCTATCACTTCTTGAGGCCAAAGATAATTTCTATGTTACTTGAAACTAG

HaplotypeSV chr1:68086419-68086450

chr1 68086443 C T 0|1

Thousand genomes (AF, EUR_AF) chr1:68086419-68086450

chr1	68086425	A	AG	0.43111	0.674	1 0
chr1	68086426	A	C	0.43111	0.674	1 0
chr1	68086428	C	T	0.43111	0.674	1 0
chr1	68086430	C	G	0.431909	0.674	1 0
chr1	68086431	T	TGG	0.43111	0.674	1 0
chr1	68086433	C	G	0.428914	0.672	1 0
chr1	68086434	ACC	A	0.43111	0.674	1 0
chr1	68086438	C	G	0.43111	0.674	1 0
chr1	68086440	A	G	0.435304	0.674	1 0
chr1	68086441	TG	T	0.43111	0.674	1 0
chr1	68086443	C	T	0.428914	0.672	1 0

PacBio chr1:68086409

68086411 68086421 68086431 68086441 68086451 68086461 68086471
AGAGCACAAAGAGATAGA*AACTCTTCACCACAATGCTCTATCACTTCTTGAGGCCAAAGATAATTTCTATGTTACTTGGA
..... M.Y.S.KSWSM.S.R..Y.....
.....GC.T.G.GGTGA.G.G.*T.....
.....gc,t,g,ggtaga,g,g,*t,.....
.....gc,t,g,ggtaga,g,g,*t,.....
.....gc,t,g,ggtaga,g,g,*t,.....
.....GC.T.G.GGTGA.G.G.TT.....
.....GC.T.G.GGTGA.G.G.*T.....
.....gc,t,g,ggtaga,g,g,*t,.....
.....*.....
.....GC.T.G.GGTGA.G.G.*T.....
.....*.....
.....*.....
.....*.....
.....*.....
.....GC.T.G.GGTGA.G.G.*T.....
.....*.....
.....*.....

Platinum & Chromium chr1:88768007-88768051

Switch process:

F1: L AGGAGAATACAGGTGATTGTGTGCTATTTTATCCAAATTAGTACTGA 1
F3: 4 AGCTTCTCCTTTCTTATGAATAAGTGCTTTCTAACTTGATGCTTAA R
RF: AGGAGAATACAGGTGATTGTGTGCTATTTTATCCAAATTAGTACTGATTGCATATTATTTAAGCATCTCTATTTTAACTACTAAGGCAAAGCTTCTCCTTTCTTATGAATAAGTGCTTTCTAACTTGATGCTTAA
RR: TCCTCTTATGTCCAATAACACACGATAAAATAGGTTTAATCATGACTAACGTATAATAAAATTCGTAGAGATAAAAATTAGTGATTCCGTTTCGAAGAGGAAGAATACTTATTCACGAAAGATTGAACACTCGAATT
F2: 3 AACGTATAATAAATTCGTAGAGATAAAAATTAGTGATTCCGTT 2

Template-switch alignment:

REF AGGAGAATACAGGTGATTGTGTGCTATTTTATCCAAATTAGTACTGA|TTGCCTTAGTGATTAAAAATAGAGATGCTTAAATAATATGCAA|AGCTTCTCCTTTCTTATGAATAAGTGCTTTCTAACTTGATGCTTAA
ALT AGGAGAATACAGGTGATTGTGTGCTATTTTATCCAAATTAGTACTGA|TTGCCTTAGTGATTAAAAATAGAGATGCTTAAATAATATGCAA|AGCTTCTCCTTTCTTATGAATAAGTGCTTTCTAACTTGATGCTTAA

Forward alignment:

REF AGGAGAATACAGGTGATTGTGTGCTATTTTATCCAAATTAGTACTGATTGcCtTAgtTgaTTAAaaATagagATgcTTAAatAaT-AtGCAAAGCTTCTCCTTTCTTATGAATAAGTGCTTTCTAACTTGATGCTTAA
ALT AGGAGAATACAGGTGATTGTGTGCTATTTTATCCAAATTAGTACTGATTG-CATATTATTTAAGCATCTCTATTTTAACTACTAAGGCCAAAGCTTCTCCTTTCTTATGAATAAGTGCTTTCTAACTTGATGCTTAA

HaplotypeSV chr1:88768007-88768051

chr1	88768010	GC	G	1 0
chr1	88768013	T	A	1 0
chr1	88768016	G	T	1 0
chr1	88768028	A	C	1 0
chr1	88768029	G	T	1 0
chr1	88768030	A	C	1 0
chr1	88768034	G	T	1 0
chr1	88768035	C	T	1 0
chr1	88768043	A	C	1 0

Switch process:
F1: L TTTCCAGGCCACCTGGCCTAGCAGAGCCACCAAGGGTGT A 1
F3: 4 CCAGAACCTCTCCCTATTATAGTTGTCATGTCAACGAGCT R

85028911 85028921 85028931 85028941 85028951 85028961 85028971
GCTTATTAAATGTGATATCCAATTTATTCACATACTGTTTACCGAAATAAAACAGTATGTG*ATAAAATGGAATATCA
.....YYKMRR.....
.....tttccg.....*

REF ATTACGATATAGACAAAAAATTAATACCTACAGCAAGTGTAAGTATAGATCagTgagaggTtAgtaagaAagGTAATATGTTTTGTTTTATCTTATTCCTATTCATAAATATGTATGAA
ALT ATTACGATATAGACAAAAAATTAATACCTACAGCAAGTGTAAGTATAGATCCTTCTTACTAAAGCTCTCACTGTAATATGTTTTGTTTTATCTTATTCCTATTCATAAATATGTATGAA

chr6	4365478	A	C	1/1
chr6	4365479	G	T	1/1
chr6	4365480	TGAGA	T	1/1
chr6	4365490	G	C	1/1
chr6	4365493	A	ACCTC	1/1

chr6	4365478	A	C	0.999201	1	1 1
chr6	4365479	G	T	0.999201	1	1 1
chr6	4365481	G	T	0.999201	1	1 1
chr6	4365490	G	C	0.999002	1	1 1
chr6	4365493	A	ACCTC	0.999002	1	1 1
chr6	4365494	G	T	0.999002	1	1 1
chr6	4365495	A	C	0.999002	1	1 1
chr6	4365497	A	C	0.999002	1	1 1
chr6	4365498	G	T	0.999002	1	1 1

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4365471 4365481 4365491 4365501 4365511 4365521
GTGTAAC TAGATACAGT GAGAGGTAGTAA****GAAGGTAATATC*TTT*GTTT*TTATCTTATTCCTATTCCATATAAT
.....CT..K.KTC...C...TC..CT.....
,,,,,,ct,****tc,,c,,cctctc,ct,,,,,,*,*,*
,,,,,,ct,****tc,,c,,cctctc,ct,,,,,,*,g,,,,,,
.....CT.****TC...C...CCTCTC.CT.....*.*
,,,,,,ct,****tc,,c,,cctctc,ct,,,,,,*,*,*
.....CT.****TC...C...CCTCTC.CT.....*.*
.....CT.****TC...C...CCTCTC.CT.....*.*
,,,,,,ct,****tc,,c,,cctctc,ct,,,,,,*,*,*
,,,,,,ct,****tc,,c,,cctctc,ct,,,,,,*,*,*
,,,,,,ct,****tc,,c,,cctctc,ct,,,,,,*,*,*
.....CT.****TC...C...CCTCTC.CT.....*.*
.....CT.****TC...C...CCTCTC.CT.....*.*
,,,,,,ct,****tc,,c,,cctctc,ct,,,,,,*,*,*
.....CT.****TC...C...CCTCTC.CT.....*.*
,,,,,,ct,****tc,,c,,cctctc,ct,,,,,,*,*,*
,,,,,,ct,****tc,,c,,cctctc,ct,,,,,,*,*,*

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F1: L TTGTATTTTAAAAATTCTTGACTGTTAAATGGCATGTTTGTGATAATT 1
 F3: 4 CTTATAGCAGATAAGTTGTAGTTAATTGGTAGTTCATGTTAAAGTTTT R
 RF: TTGTATTTTAAAAATTCTTGACTGTTAAATGGCATGTTTGTGATAATTAGGAAAAAGAAGCAAGATATTAGTCTTATAGCAGATAAGTTGTAGTTAATTGGTAGTTCATGTTAAAGTTTT
 RR: AACATAAAATTTTAAAGAACTGACAATTTTACCGTACAAACACTATTAATCTCTTTTCTCGTTCTATATAATCGAGAATAATCGTCTATTCAACATCAATTAAACCATCAAGTACAATTTCAAAA
 F2: 3 TCCTTTTCTCGTTCTATATAATCGA 2

REF TTGTAATTTAAAAATCTTGACTGTTAAAAATGGCATGTTTGTGATAAAT|AGCTAATATATCTTGCTTCTTTTTCCT|CTTATAGCAGATAAGTGTAGTTAATTGGTAGTTCATGTTAAAGTTTT
ALT TTGTAATTTAAAAATCTTGACTGTTAAAAATGGCATGTTTGTGATAAAT|AGCTAATATATCTTGCTTCTTTTTCCT|CTTATAGCAGATAAGTGTAGTTAATTGGTAGTTCATGTTAAAGTTTT

REF TTGTAATTTTAAAAAATCTTGACTGTGTAATAATGGCATGTTTGTGATAATTAGTcAtAatAtAtccttgctTcTtTtctCTCTTATAGCAGATAAGTTGTAGTTAATTGGTAGTTCATGTTAAAGTTTT
ALT TTGTAATTTTAAAAAATCTTGACTGTGTAATAATGGCATGTTTGTGATAATTAGGAAAAAGAAGCAAGATATATTAGCTCTTATAGCAGATAAGTTGTAGTTAATTGGTAGTTCATGTTAAAGTTTT

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chr6 20824935 T A 0|1
chr6 20824945 T A 0|1
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chr6	20824934	C	G	0.358826	0.327	0 1
chr6	20824935	T	A	0.358826	0.327	0 1
chr6	20824938	T	A	0.326478	0.2853	0 0
chr6	20824939	A	AG	0.358826	0.327	0 1
chr6	20824940	T	A	0.358826	0.327	0 1
chr6	20824942	T	G	0.358826	0.327	0 1
chr6	20824944	T	A	0.358826	0.327	0 1
chr6	20824945	T	A	0.326478	0.2853	0 0
chr6	20824946	GC	G	0.358826	0.327	0 1
chr6	20824948	T	A	0.358826	0.327	0 1
chr6	20824950	C	A	0.358826	0.327	0 1
chr6	20824952	T	A	0.358826	0.327	0 1
chr6	20824955	T	A	0.358826	0.327	0 1

Switch process:
F1: L GTGATACGTTGGCTTGGCCATTTCAGACTCTCTGTCTGCATTTCTGTGTCT 1
F3: 4 TTTTCGACCACTGCCAGGGGGCGGATTATTTTGGATTCTTTCCTCCAA R
RF: GTGATACGTTGGCTTGGCCATTTCAGACTCTCTGTCTGCATTTCTGTGTCTTTGCACTATTAATCAACAGCTCACCCATCAATTTTCGACCACTGCCAGGGGGCGGATTATTTTGGATTCTTTCCTCCAA
RR: CACTATCGAACCGAACCAGTAACCGTCAGAGACAGACGTAAGACACAGAAACGTGATAATTAGTTGTGCGAGTGGGTAGTTAAAGCTGGTCACGCTCCCCCGCTAATAAAACCTAAGAAAGGAGGTT

F2: 3 AACGTGATAATTAGTTGTCGAGTGGGTAGTT 2

Template-switch alignment:

REF GTGATACGTGGCTTGGCCATTGTCAGTCTCTGTCTGCATTCTGTGTCT TTGATGGGTGAGCTGTTGATTAATAGTGCAA TTTTCGACCAGTGCCAGGGGGCGGATTATTTGGATTCTTCTCCAA
ALT GTGATACGTGGCTTGGCCATTGTCAGTCTCTGTCTGCATTCTGTGTCT TTGATGGGTGAGCTGTTGATTAATAGTGCAA TTTTCGACCAGTGCCAGGGGGCGGATTATTTGGATTCTTCTCCAA

Forward alignment:

REF GTGATACGTTGGCTTGGCCATTTCAGTCTCTGTCTGCATTTCGTGTCTTtgatgggTgAgCTgTtGAtTAAtAgT-----gCAATTTTCGACCAGTGCCAGGGGGCGATTATTTTGGATTCTTCTCCAA
ALT GTGATACGTTGGCTTGGCCATTTCAGTCTCTGTCTGCATTTCGTGTCTT-----TGCACtTAATCAACAGCtCACCCATCAATTTTCGACCAGTGCCAGGGGGCGATTATTTTGGATTCTTCTCCAA

HaplotypeSV chr7:28419452-28419484

NA

Thousand genomes (AF, EUR_AF) chr7:28419452-28419484

NA

PacBio chr7:28419442

[illegible]

— — —

Platinum & Chromium chr7:117716979-117717015

Switch process:

F1: L TTATAATCCTGCTCTCTGCGCCCCACCTTCCACCCACACACAGGTCT 1

F1: T TATAATCTCGTCTCTCTCGCGCCCCACCTTCCCACCCACACACAGGTCTTT
F2: 4 TCAATCAACATGGTTTGGTGAACAAACCATATATTACAAAGTGCTTC R
RF: TTATAATCTCGTCTCTCTCGCGCCCCACCTTCCCACCCACACACAGGTCTTTGTAAGCCACCTTGGGAGTAGATGAGAACGGCAATCAATCAACATGGTTTGGTGAACAAACCATATATTACAAAGTGCTTC
RR: AATATTAGGACGAGAGACGCGGGGTGGAAGGGTGGGTGTGTGCCAACCAATTCGGTGGAAACCCCTCATCTACTCTTGGCGGTTAGTTAGTTGTACCAAAACCATCTGTTTGGGTATATAATCTTTCACGAAG
F2: 3 AAACATCTCGGTGGAACCCCTGATCTACTCTTGGCGGT 2

Template-switch alignment:

Complete match alignment:

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REF  TTATAATCTGCTGCTCTGCGGCCCACTTCCACCCACACACAGGTCT  TTGCGGTTCTCATCTACTCCCAAGTGGGTTACAA  TCAATCAACATGGTTTGTGGAACAAACCATATATTACAAAGTGCTTC
ALT  TTATAATCTGCTGCTCTGCGGCCCACTTCCACCCACACACAGGTCT  TTGCGGTTCTCATCTACTCCCAAGTGGGTTACAA  TCAATCAACATGGTTTGTGGAACAAACCATATATTACAAAGTGCTTC
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Forward alignment:

REF TTATAATCTCTGCTCTCTGCGCCCCACCTTCCCAACCCACACAGGTCCTTTCgcttCtCaTctactcccaAgTgGgttaCAATCAATCAACATGGTTTGGTGAACAAACCATATATTACAAAGTGCTTC
ALT TTATAATCTCTGCTCTCTGCGCCCCACCTTCCCAACCCACACAGGTCCTTGTGAAGCCACCTTGGGAGTAGATGAGAACGGCAATCAATCAACATGGTTTGGTGAACAAACCATATATTACAAAGTGCTTC

HaplotypeSV chr7:117716979-117717015

NA

Thousand genomes (AF, EUR_AF) chr7:117716979-117717015

NA

PacBio chr7:117716969

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117716971 117716961 117716991 117717001 117717011 117717021 117717031
CACACAGGTGCTTTGGCGTTCATCATCACTCCCAAG*GTG+G*CTTACAATCAATCAACATGTTTGGTGAAACAACCAT
.....KTAAG...C..TGGGAGT..A...ACGG.....
.....*TAAG*..C..TGGGAGT*..A..AACGG.....
,,,,,,,,,,,,,*taag*,c,,tggggagt,*,a,,a,aacgg,,,,,,,,,,,,
,,,,,,,,,,,,,*TAAG*..C..TGGGAGT*..A..AACGG.....
,,,,,,,,,,,,,*taag*,c,,tgggagt,*,a,,a,aacgg,,,,,,,,,,,,
,,,,,,,,,,,,,*TAAG*..C..TGGGAGT*..A..AACGG.....
,,,,,,,,,,,,,*taag*,c,,tggggagt,*,a,,a,aacgg,,,,,,,,,,,,*
,,,,,,,,,,,,,*TAAG*..C..TGGGAGT*..A..AACGG.....
,,,,,,,,,,,,,*taag*,c,,tgggagt,*,a,,a,aacgg,,,,,,,,,,,,
,,,,,,,,,,,,,*TAAG*..C..TGGGAGT*..A..AACGG.....
,,,,,,,,,,,,,*taag*,c,,tggggagt,*,a,,a,aacgg,,,,,,,,,,,,
,,,,,,,,,,,,,*TAAG*..C..TGGGAGT*..A..*A*ACGG.....
,,,,,,,,,,,,,*TAAG*..C..TGGGAGT*..A..AACGG.....
,,,,,,,,,,,,,*taag*,c,,tgggagt,*,a,,a,aacgg,,,,,,,,,,,,

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,,,,,,*taag*,c,gggagt,,a,,a,aacgg,,,,,,,,,,,,,
,,,,,,*taag*,c,gggagt,,a,,a,aacgg,,,,,,,,,,,,,
.....*TAAG*..C..TGGGAGT..*A..A.AACGG.....
.....*TAAG*..C..TGGGAGT..*A..A.AACGG.....

Platinum & Chromium chr8:31526297-31526340

Switch process:

F1: L ATATTTAAACATATAAATATTTTAAACACATAAATGTCCACTGCT 1
F3: 4 GACTGCAAATGGAGTAGAGCTGATTGTAACATTATATGCAAATGAG R
RF: ATATTTAAACATATAAATATTTTAAACACATAAATGTCCACTGCTAATTGCCTGGCAATTTGACTGCAAATGGAGTAGAGCTGATTGTAACATTATATGCAAATGAG
RR: TATAAATTTGTATATTATATAAATTTGTGTATTACAGGTGACGATTAACGGACCGTTAAACTGACGTTTACCTCATCTCGACTAACATTGTAATATACGTTTACT
F2: 3 TTTACAGGTGACGATTAACGGACCGTTAA 2

Template-switch alignment:

REF ATATTTAAACATATAAATATTTTAAACACATAAATGTCCACTGCT|AATTGCCAGGCAATTAGCAGTGGACATTT|GACTGCAAATGGAGTAGAGCTGATTGTAACATTATATGCAAATGAG
ALT ATATTTAAACATATAAATATTTTAAACACATAAATGTCCACTGCT|AATTGCCAGGCAATTAGCAGTGGACATTT|GACTGCAAATGGAGTAGAGCTGATTGTAACATTATATGCAAATGAG

Forward alignment:

REF ATATTTAAACATATAAATATTTTAAACACATAAATGTCCACTGCTAATTGCCaGGCAattagcagtggacATTTGACTGCAAATGGAGTAGAGCTGATTGTAACATTATATGCAAATGAG
ALT ATATTTAAACATATAAATATTTTAAACACATAAATGTCCACTGCTAATTGCCCTGGCA-----ATTGACTGCAAATGGAGTAGAGCTGATTGTAACATTATATGCAAATGAG

HaplotypeSV chr8:31526297-31526340

NA

Thousand genomes (AF, EUR_AF) chr8:31526297-31526340

chr8	31526319	A	T	0.0061901	0.0209	0 1
chr8	31526323	AATTAGCAGTGGAC	A	0.00599042	0.0209	0 1

PacBio chr8:31526287

31526291 31526301 31526311 31526321 31526331 31526341 31526351
TTT*AAACACATAAATGTCCACTGCTAATTGCCAGGCAATTAGCAGTGGACATTTGACTGCAAATGGAGTAGAGCTGATT
.....W.....
,,*,,,,,,,a,,,,,
,,*,,,,,,,
,,*,,,,,,,
,,*,,,,,,,t,,,,*****
..A.....T.....*****
,,*,,,,,,,t,,,,*****
,,*,,,,,,,
..*.....
..*.....
,,*,,,,,,,
,,*,,,,,,,
,,*,,,,,,,
,,*,,,,,,,t,,,,*****
,,a*,,,,,,,
,,*,,,,,,,
,,*,,,,,,,t,,,,*****
,,*,,,,,,,t,,,,*****

Platinum & Chromium chr10:3134417-3134449

Switch process:

F1: L TATAAGGACCTAGAAATAAAAAACATGAATTCGCAAATAGAAATTGTC 1
F3: 4 AACTGGTATTTTCATATAACTCTAACCACCAACAGGGTGGGGCACCCCTC R
RF: TATAAGGACCTAGAAATAAAAAACATGAATTCGCAAATAGAAATTGTCATACAGTAACACTTTGGTGTTAAATAGAAATAACTGGTATTTTCATATAACTCTAACCACCAACAGGGTGGGGCACCCCTC
RR: ATATTCTGGATCTTTATTTTGTACTTAAGACGTTTTATCTTTAACAGTATGTCAATTGTGAAACGACAATTTATCTTTATTGACCATAAAGTATATTGAGATTGGTGGTGTGCCACCCCGTGGGAG
F2: 3 TATGTCAATTGTGAAACGACAATTTATCTTTA 2

Template-switch alignment:

REF TATAAGGACCTAGAAATAAAAAACATGAATTCGCAAATAGAAATTGTC|ATTTCTATTTAACAGCAAAGTGTTACTGTAT|AACTGGTATTTTCATATAACTCTAACCACCAACAGGGTGGGGCACCCCTC
ALT TATAAGGACCTAGAAATAAAAAACATGAATTCGCAAATAGAAATTGTC|ATTTCTATTTAACAGCAAAGTGTTACTGTAT|AACTGGTATTTTCATATAACTCTAACCACCAACAGGGTGGGGCACCCCTC

Forward alignment:

REF TATAAGGACCTAGAAATAAAAAACATGAATTCGCAAATAGAAATTGTCATtctattttaACAGcAAagtgTTaCTGT-----ATAACTGGTATTTTCATATAACTCTAACCACCAACAGGGTGGGGCACCCCTC
ALT TATAAGGACCTAGAAATAAAAAACATGAATTCGCAAATAGAAATTGTCAT-----ACAGTAACACTTTGTGTTAAATAGAAATAACTGGTATTTTCATATAACTCTAACCACCAACAGGGTGGGGCACCCCTC

HaplotypeSV chr10:3134417-3134449

chr10	3134418	AT	A	1 0
chr10	3134422	CT	C	1 0
chr10	3134434	A	T	1 0
chr10	3134441	T	TA	1 0

Thousand genomes (AF, EUR_AF) chr10:3134417-3134449

chr10	3134419	TTTCTATTTA	T	0.086262	0.1193	1 0
chr10	3134433	C	T	0.0966454	0.1193	1 0
chr10	3134435	A	AC	0.106829	0.1064	0 0

F1: L CATAGGACTCCTAATAGAAAAAGTGATTCATAGCAAAATAATG 1
F3: 4 AGAAGATAGACACTAGTCAATGATCAGTTTCATATTATCATAAAAAGTAA R
RF: CATAGGACTCCTAATAGAAAAAGTGATTCATAGCAAAATAATGTTTCATAGCCTTTGCTATGAAAGAAAGATAGACACTAGTCAATGATCAGTTTCATATTATCATAAAAAGTAA
RR: GTATCCTCGAGGATTATCTTTTCACTAAGTATCGTTTTATTACAAAGATATCGGAAACGATACTTTCTTCTATCTGTGATCAGTTACTAGTCAAAGTATAATAGTATTTTTCATT
F2: 3 CACTTTTATAACAAAGTATCGGAAACGATACTTT 2

ALT CATAGGACTCCTAATAGAAAAGTGATTATAGCAAAAATATG|TTTCATAGCAAAGGCTATGAACATTATTTcaC|AGAAGATAGACACTAGTCAATGATCAGTTTCATATTATCATAAAAAGTAA
REF CATAGGACTCCTAATAGAAAAGTGATTATAGCAAAAATATG|TTTCATAGCAAAGGCTATGAACATTATTTTGC|AGAAGATAGACACTAGTCAATGATCAGTTTCATATTATCATAAAAAGTAA

ALT CATAGGACTCCTAATAGAAAAGTGATTATAGCAAAAATAATGTTTCATAGCAaagGCTATGAacattatttcacAGAAGATAGACACTAGTCAATGATCAGTTTCATATTATCAAAAAAGTAA
REF CATAGGACTCCTAATAGAAAAGTGATTATAGCAAAAATAATGTTTCATAGCCTTGTCTATGAA-----AGAAGATAGACACTAGTCAATGATCAGTTTCATATTATCAAAAAAGTAA

chr12 133076355 T A 1|1

NA

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133076331 133076341 133076351 133076361 133076371 133076381
GATTTCATAGCAAAATAAT*GTTTCATAGCCTTGGCTATGAA*****AGAAGATAGACACTAGTCAATGATTCAG
.....AAAG.....
.....*.....,aaag,,,,,,acattatttcac,.....
.....*.....,aaag,,,,,,acattatttcac,.....
.....*.....,aaag,,,,,,acattatttcac,.....
.....*......AAAG......ACATTATTTCAC.
.....*.....,aaag,,,,,,acattatttcac,.....
.....*.....,aaag,,,,,,acattatttcac,.....
.....*.....,aaag,,,,,,acattatttcac,.....
.....*......AAAG......ACATTATTTCAC.
.....*.....,aaag,,,,,,acattatttcac,.....
.....*.....,aaag,,,,,,acattatttcac,.....
.....*.....,aaag,,,,,,acattatttcac,.....
.....*......AAAG......ACATTATTTCAC.
.....*......AAAG......ACATTATTTCAC.
.....*.....,aaag,,,,,,acattatttcac,.....
.....*.....,aaag,,,,,,acattatttcac,.....
.....*......AAAG......ACATTATTTCAC.

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Platinum & Chromium chr17:61667231-61667263

F1: L TAAAGATATTATCATTAAATTCATTACCACCACCAAGTACTGCTTCTCA 1
F3: 4 AGAATGTGACTGAGTGGCTTTTTTTTGGAGAGACTTGTCTGTGTC R
RF: TAAAGATATTATCATTAAATTCATTACCACCACCAAGTACTGCTTCTCAATAATTCATGTTACATGCAATCACAAAGAAATGTGACTGAGTGGCTTTTTTTTGGAGAGACTTGTCTGTGTC
RR: ATTTCTATAATAGTAATTTAAGATAAATGGTGGTGGTTCATGACGAAGAGTAATATTTAAGATACCAATGTACGTTAGTGTCTTTCACACTGACTCACCAGAAAAAAACTCTCTCAGAACGAGACAACG
F2: 3 AATAATTGAATACCAATGTACGTTAGTGT 2

REF TAAAGATATTATCATTAATAATCTATTACCACCACCAAGTACTGCTTCTCA|TTGTGATTGCATGTAACCATAGAATTATAA|AGAATGTGACTGAGTGGCTTTTTTTTTTGAGAGAGTCTTGCTCTGTTGC
ALT TAAAGATATTATCATTAATAATCTATTACCACCACCAAGTACTGCTTCTCA|TTGTGATTGCATGTAACCATAGAATTATAA|AGAATGTGACTGAGTGGCTTTTTTTTTTGAGAGAGTCTTGCTCTGTTGC

REF TAAAGATATTATCATTTAAATTCATTACCACCACCAAGTACTGCTTCTCAT-----TgTgATgCATGtAAccatagaatTtAtAAAGAAATGTGACTGAGTGGCTTTTTTTTTTGAGAGAGTCTTGCTCTGTTCG
ALT TAAAGATATTATCATTTAAATTCATTACCACCACCAAGTACTGCTTCTCATTATAAATTCATGGTTACATGCAA-----TCACAAAGAAATGTGACTGAGTGGCTTTTTTTTTTGAGAGAGTCTTGCTCTGTTCG

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chr17 61667234 G A 1|1
chr17 61667236 G A 1|1
chr17 61667260 T C 1|1
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chr17	61667234	G	A	0.564297	0.4761	1 1
chr17	61667241	C	T	0.563099	0.4751	1 1
chr17	61667254	A	C	0.563099	0.4751	1 1
chr17	61667256	T	A	0.563099	0.4751	1 1
chr17	61667258	T	C	0.563299	0.4751	1 1
chr17	61667260	T	C	0.563299	0.4751	1 1

[illegible]

,,,,,,,,,a,aa,,ct,,g,,t,,,*,*,c,a,c,c,,,,,,,,,,,,,,,,,,,,,,,,,,,,,
,,,,,,,,,a,aa,,ct,,g,,t,,,*,*,c,a,c,c,,,,,,,,,,,,,,,,,,,,,,,,,,,,,
g,,,,,,,,,a,aa,,ct,,g,,t,,,*,*,c,a,c,c,,,,,,,,,,,,,,,,,,,,,,,,,,,,,
g,,,,,,,,,a,aa,,ct,,g,,t,,,*,*,c,a,c,c,,,,,,,,,,,,,,,,,,,,,,,,,,,,,
.G.....A.AA...CT..G..T.*...*.C.A.C.C.....
,,,,,,,,,a,aa,,ct,,g,,t,,,*,*,c,a,c,c,,,,,,,,,,,,,,,,,,,,,,,,,,,,,
.....A.AA...CT..G..T.*...*.C.A.C.C.....
,,,,,,,,,a,aa,,ct,,g,,t,,,*,*,c,a,c,c,,,,,,,,,,,,,,,,,,,,,,,,,,,,,
.....A.AA...CT..G..T.*...*.C.A.C.C.....
.....A.AA...CT..G..T.*...*.C.A.C.C.....