

Supplementary File 1

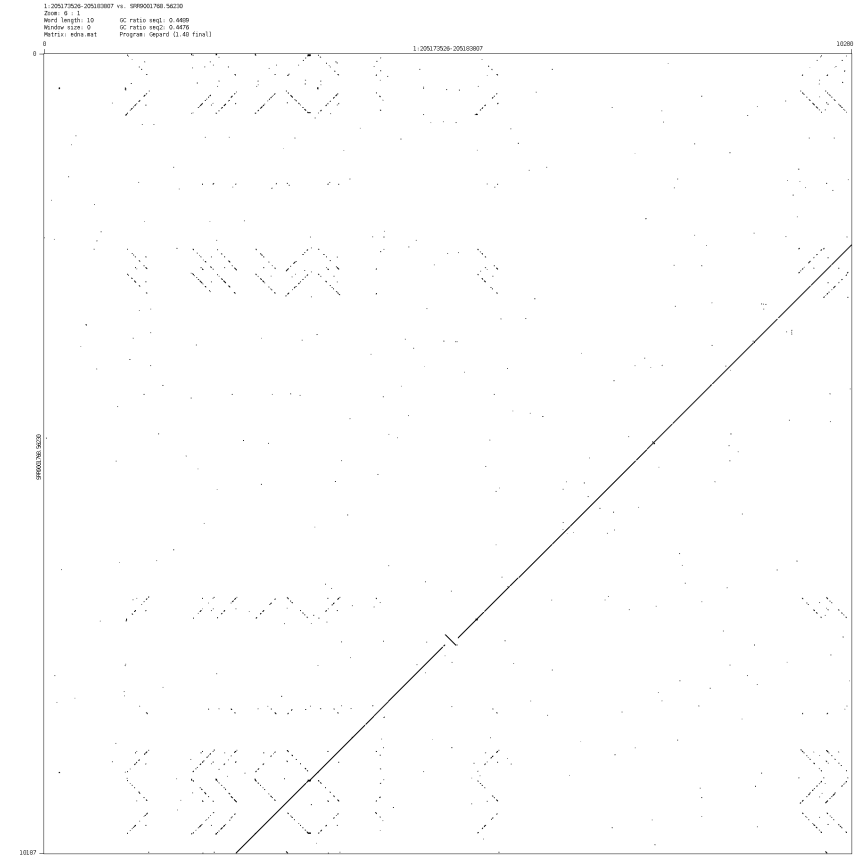
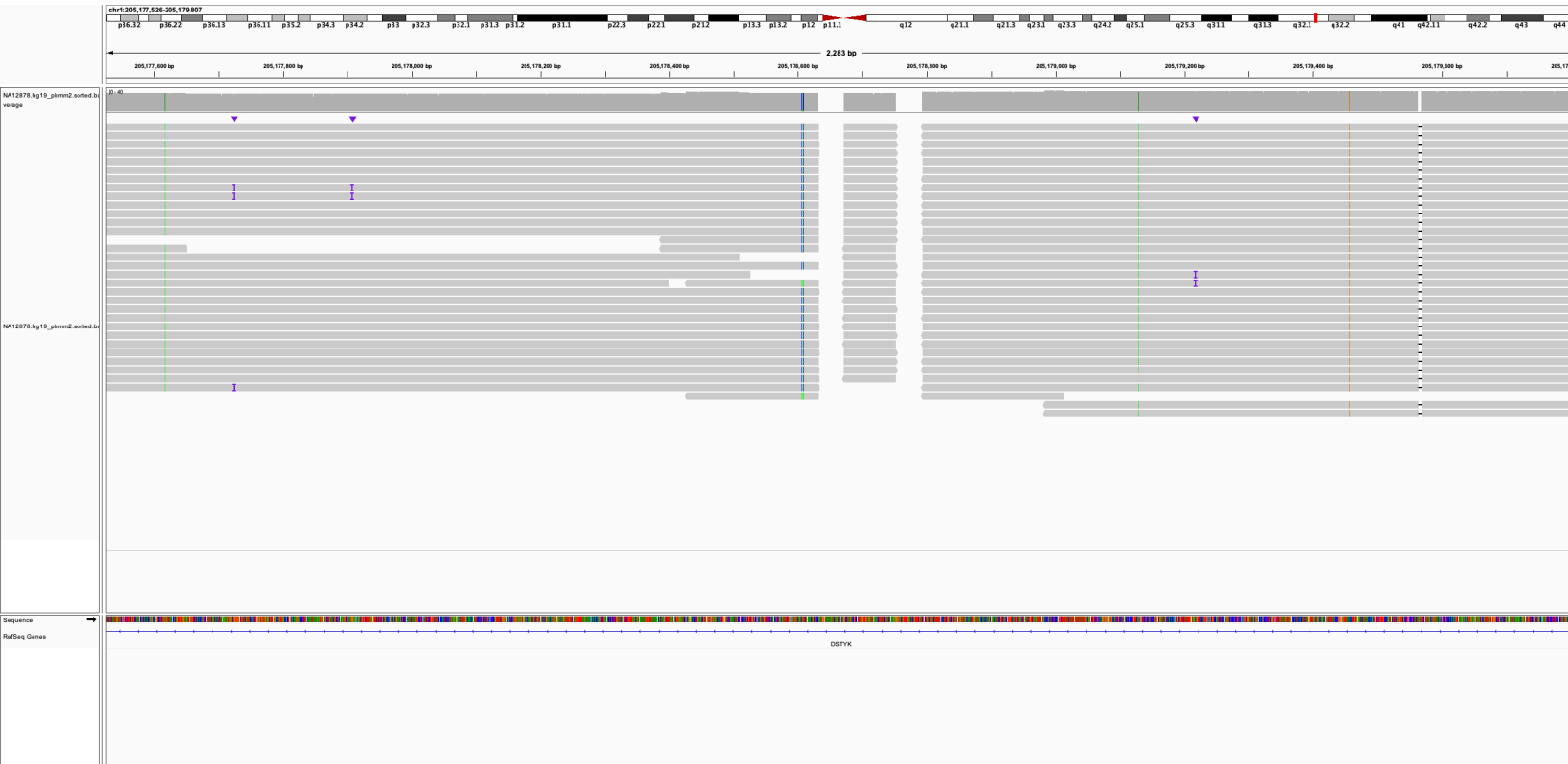
Manually curated CSV loci in NA12878 initially reported by 1000 Genomes Project

Note:

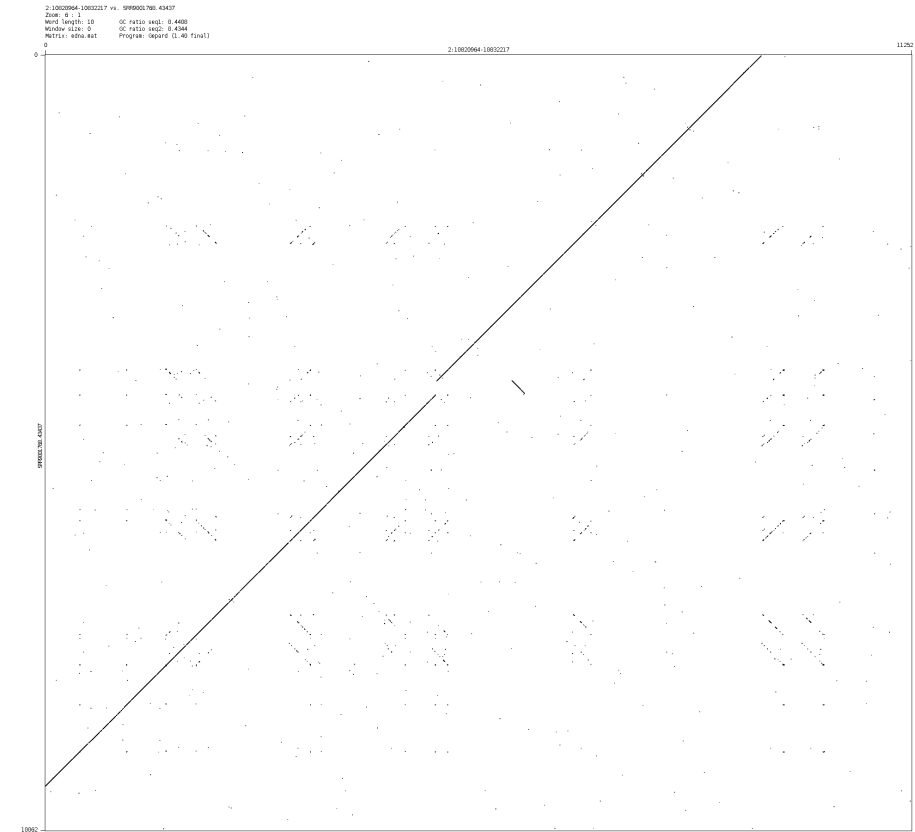
- The IGV screenshots and Dotplots for 18 curated CSV and 64 simple SVs are listed here.
- The raw Dotplots of 82 true discoveries and 231 false discoveries are available at: <https://drive.google.com/file/d/1YjuQOpjJ0JSPk9nq1GA7oQxjF6ii6dR0/view?usp=sharing>.
- The results in this file are summarized in Supplementary Table S5.

Complex structural variants

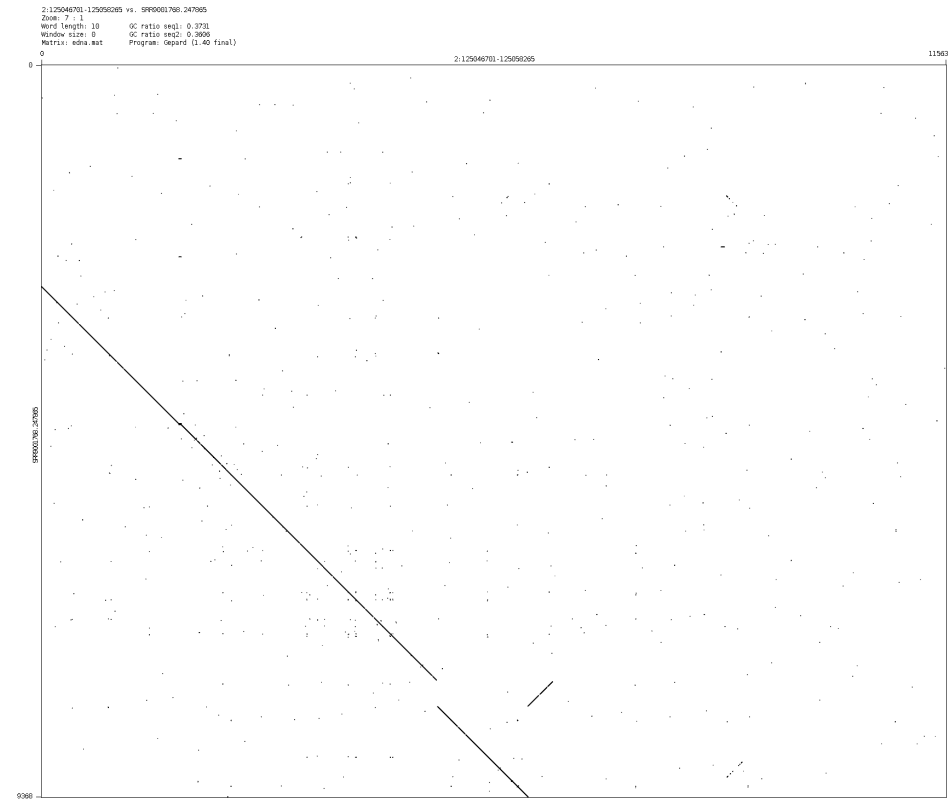
❖ chr1:205,178,526-205,178,807



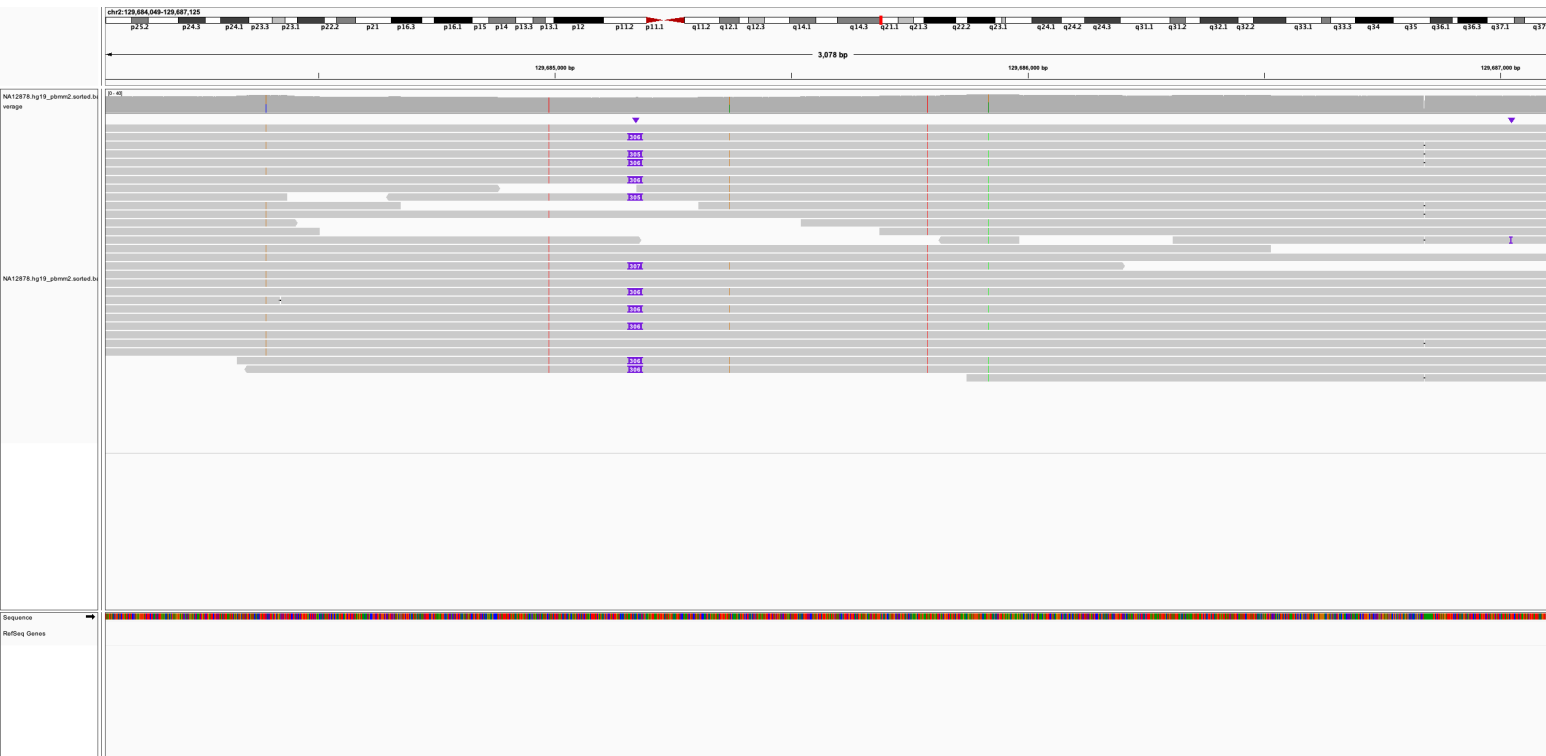
❖ chr2:10,825,964-10,827,217



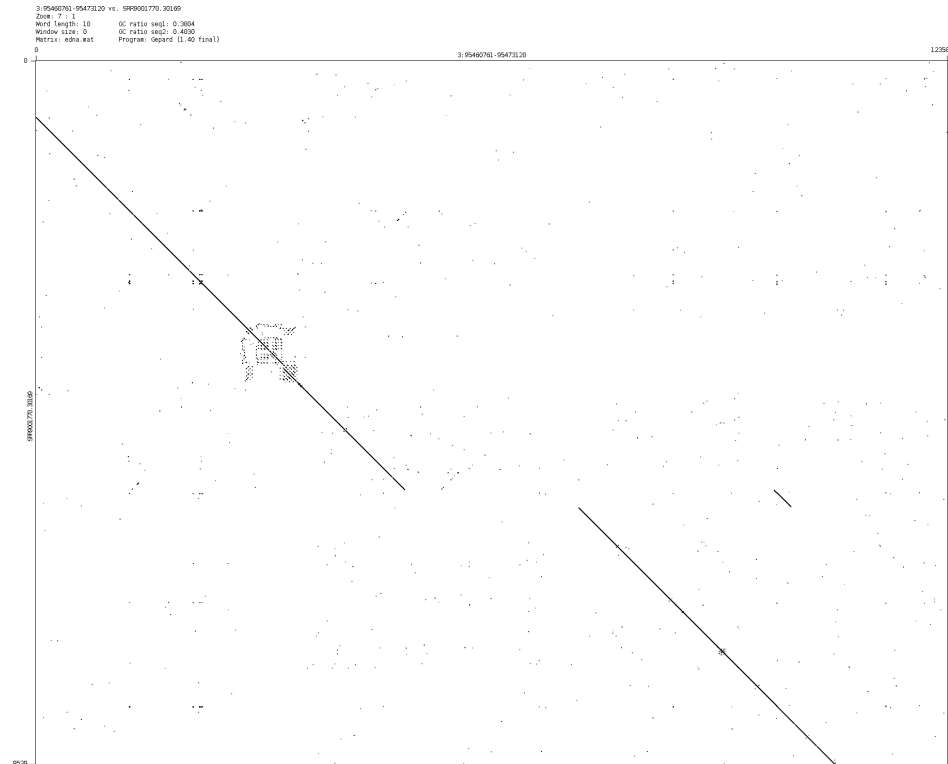
❖ chr2:125,051,701-125,053,265



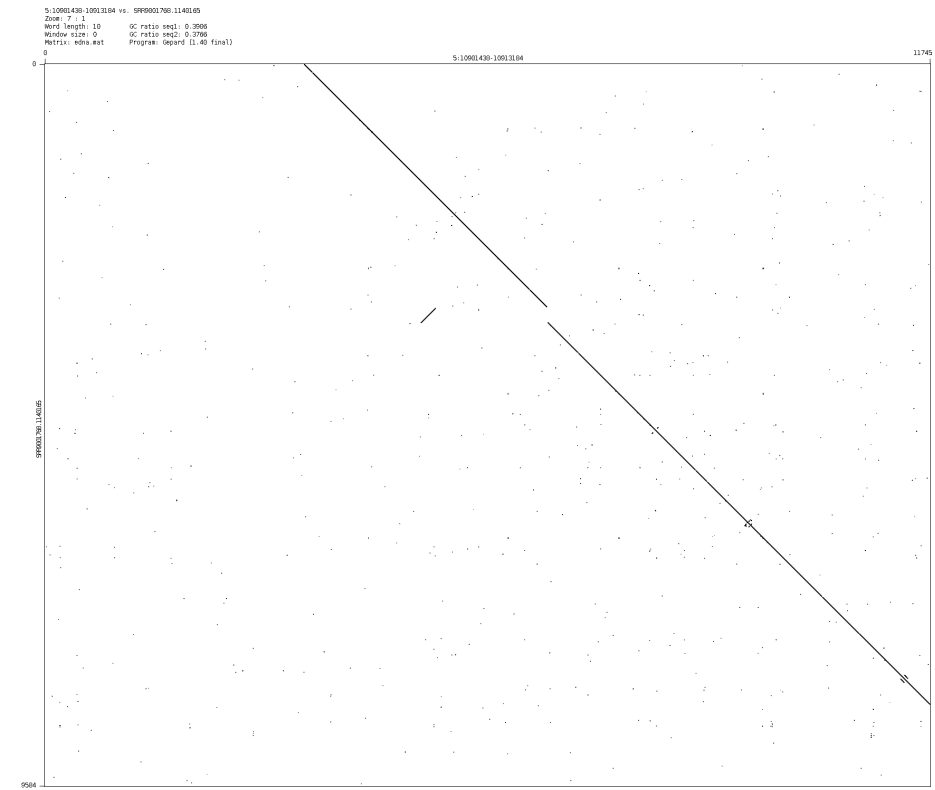
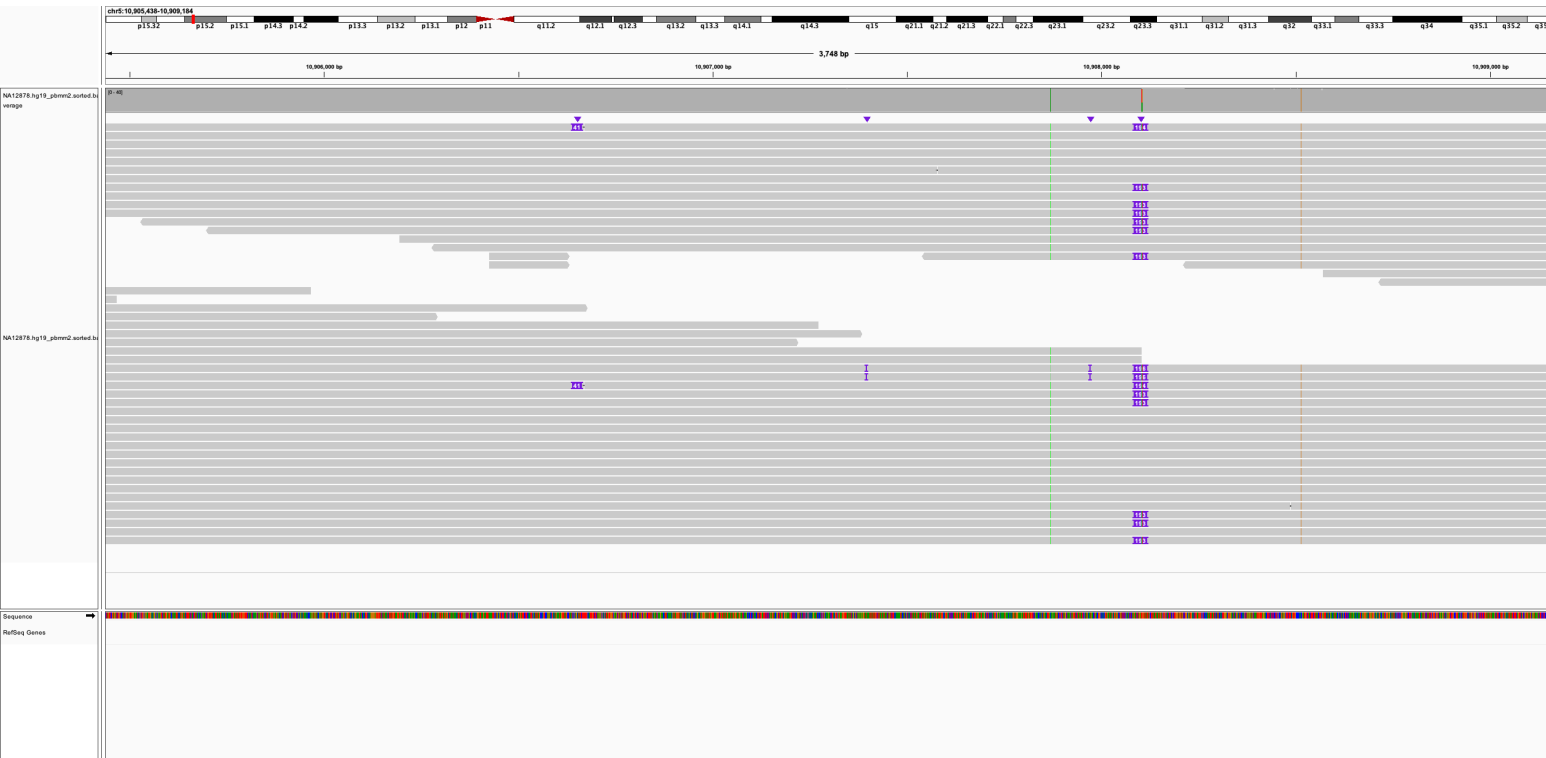
❖ chr2:129,685,049-129,686,125



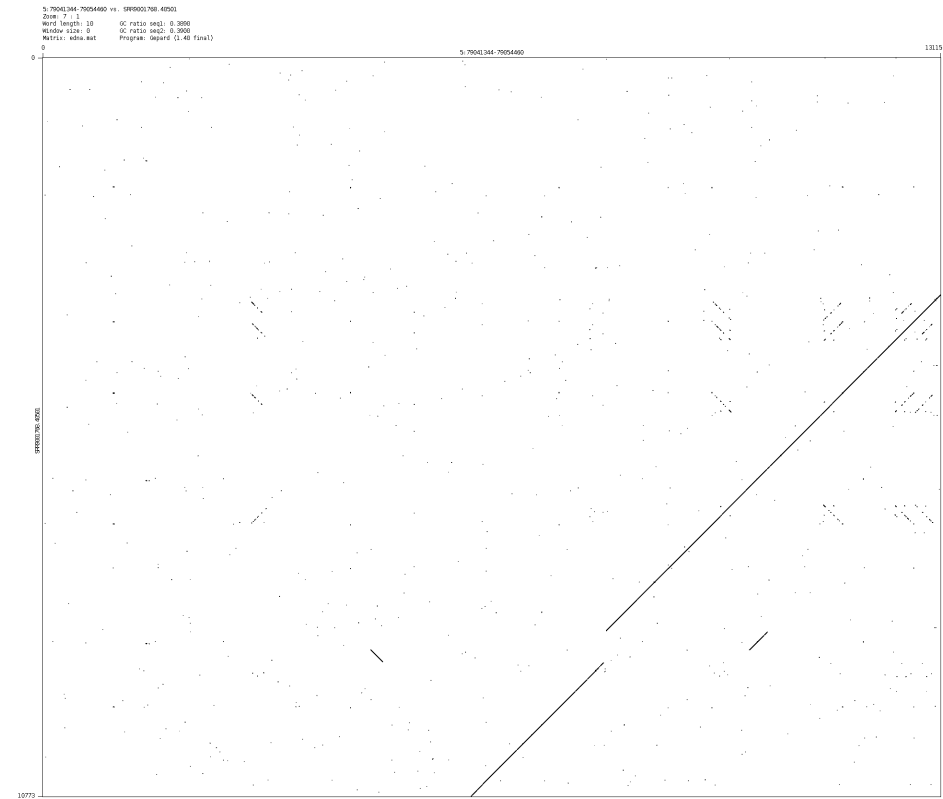
❖ chr3:95,465,761-95,468,120



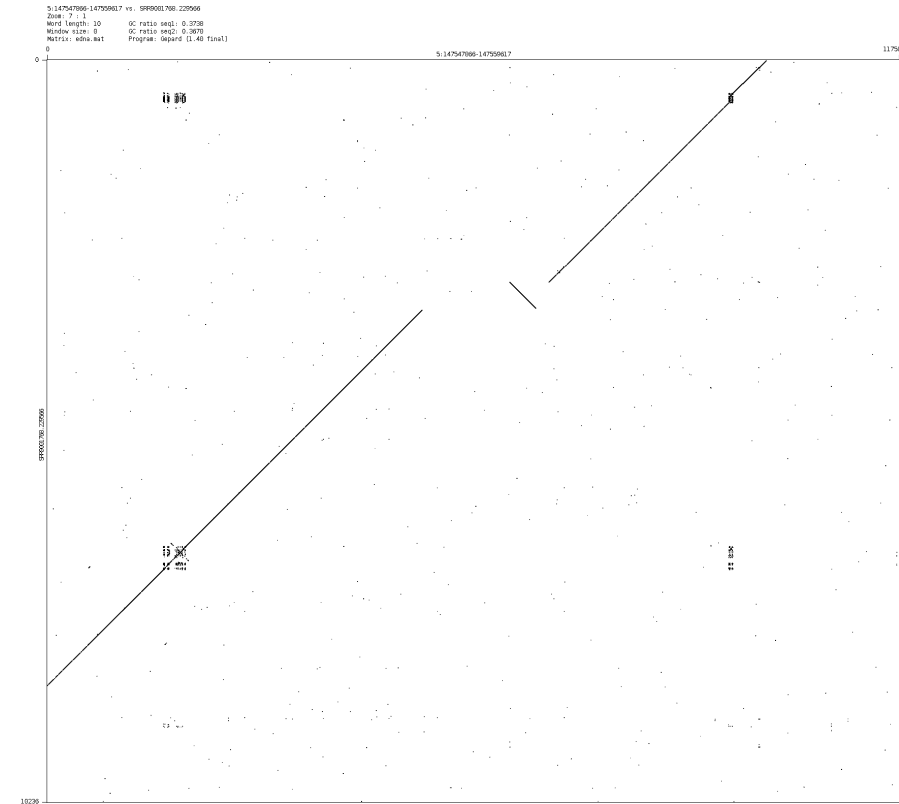
❖ chr5:10,906,438-10,908,184



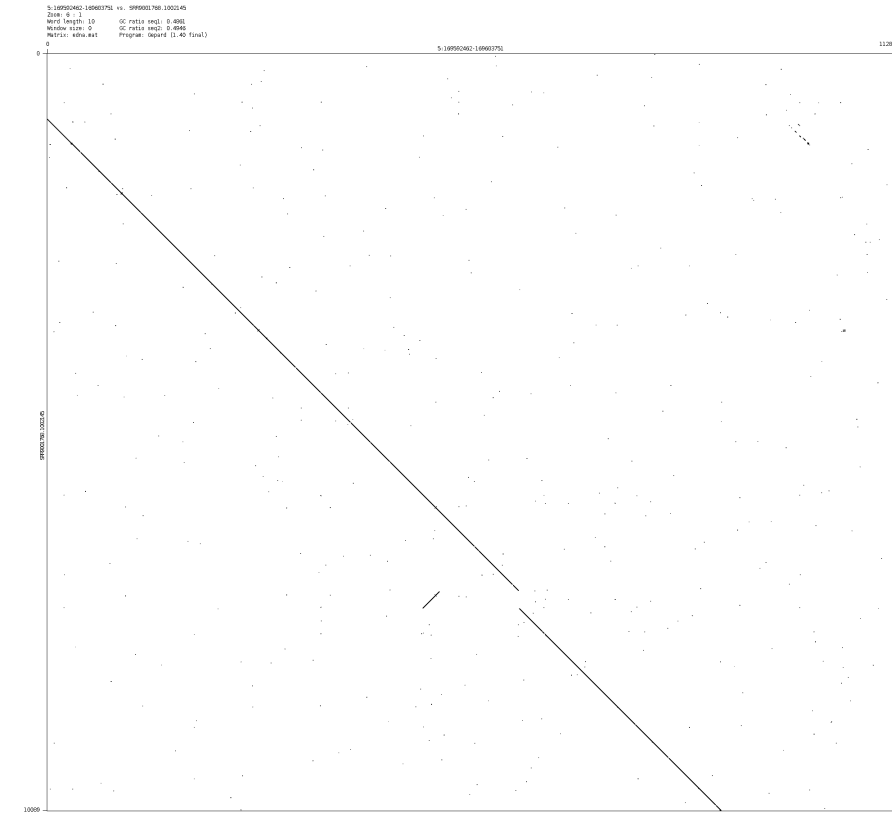
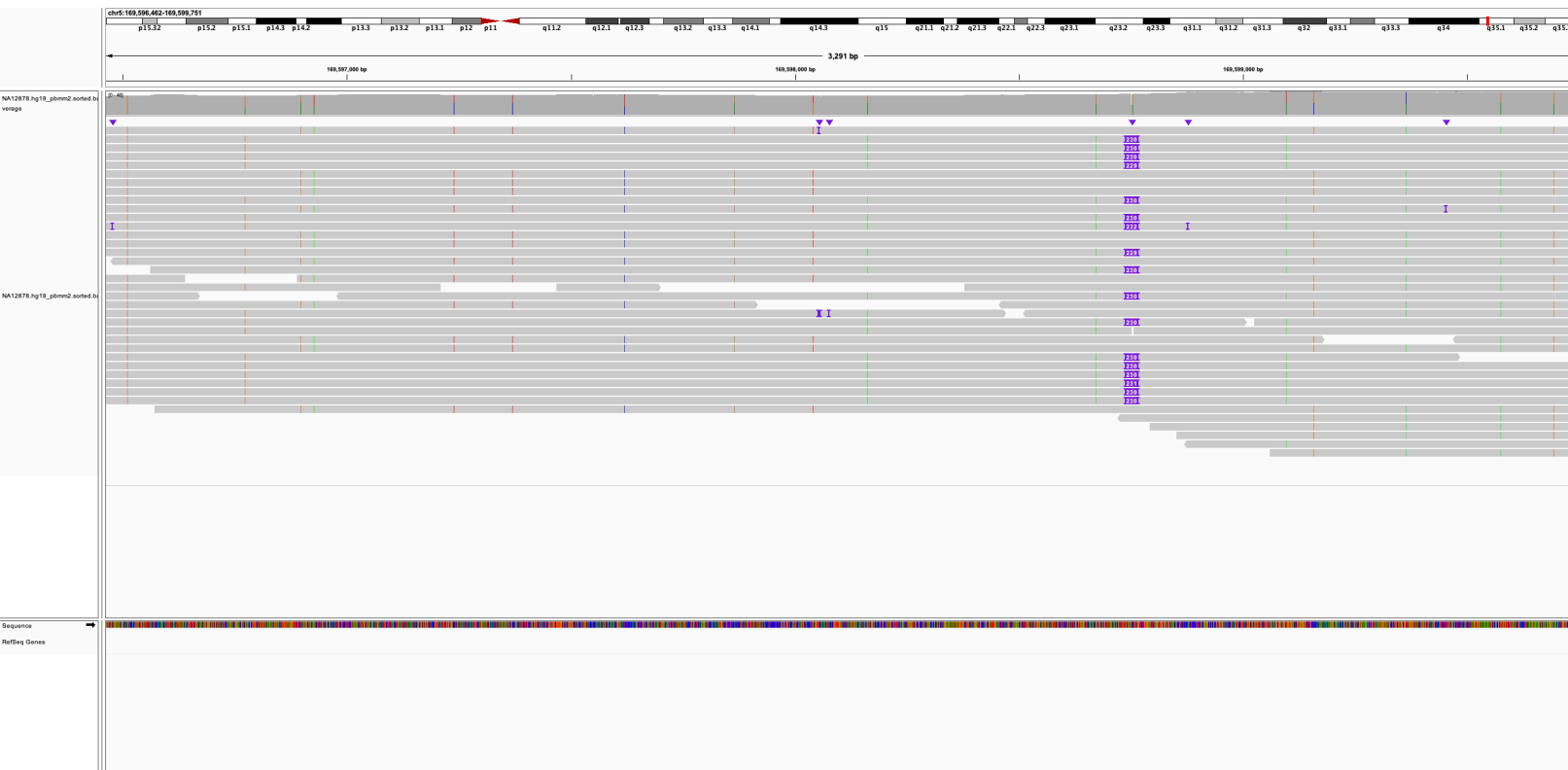
❖ chr5:79,046,344-79,049,460



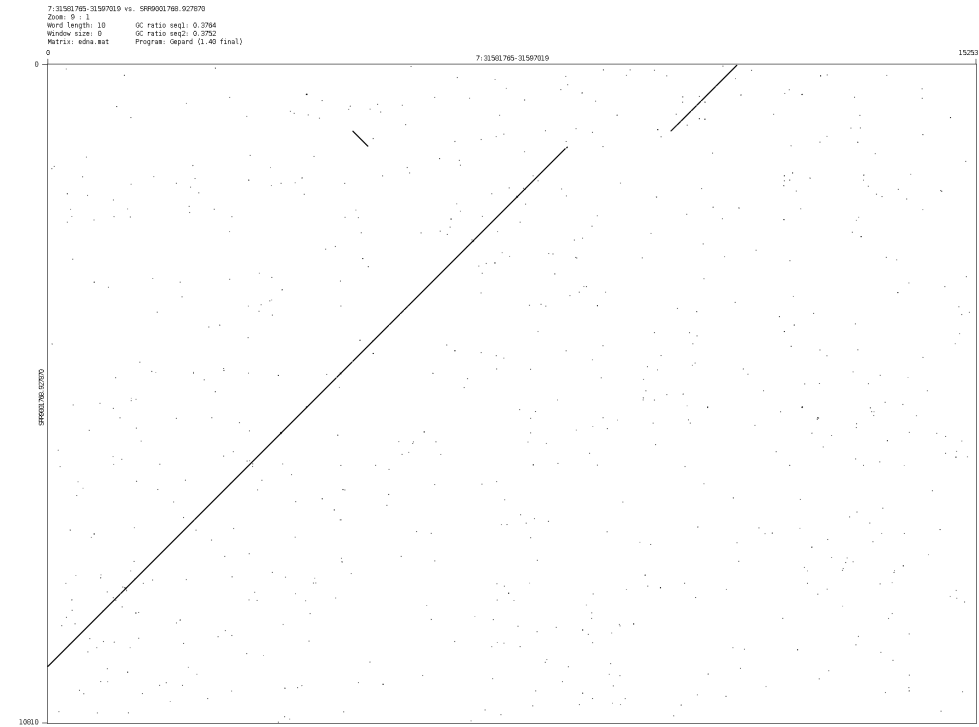
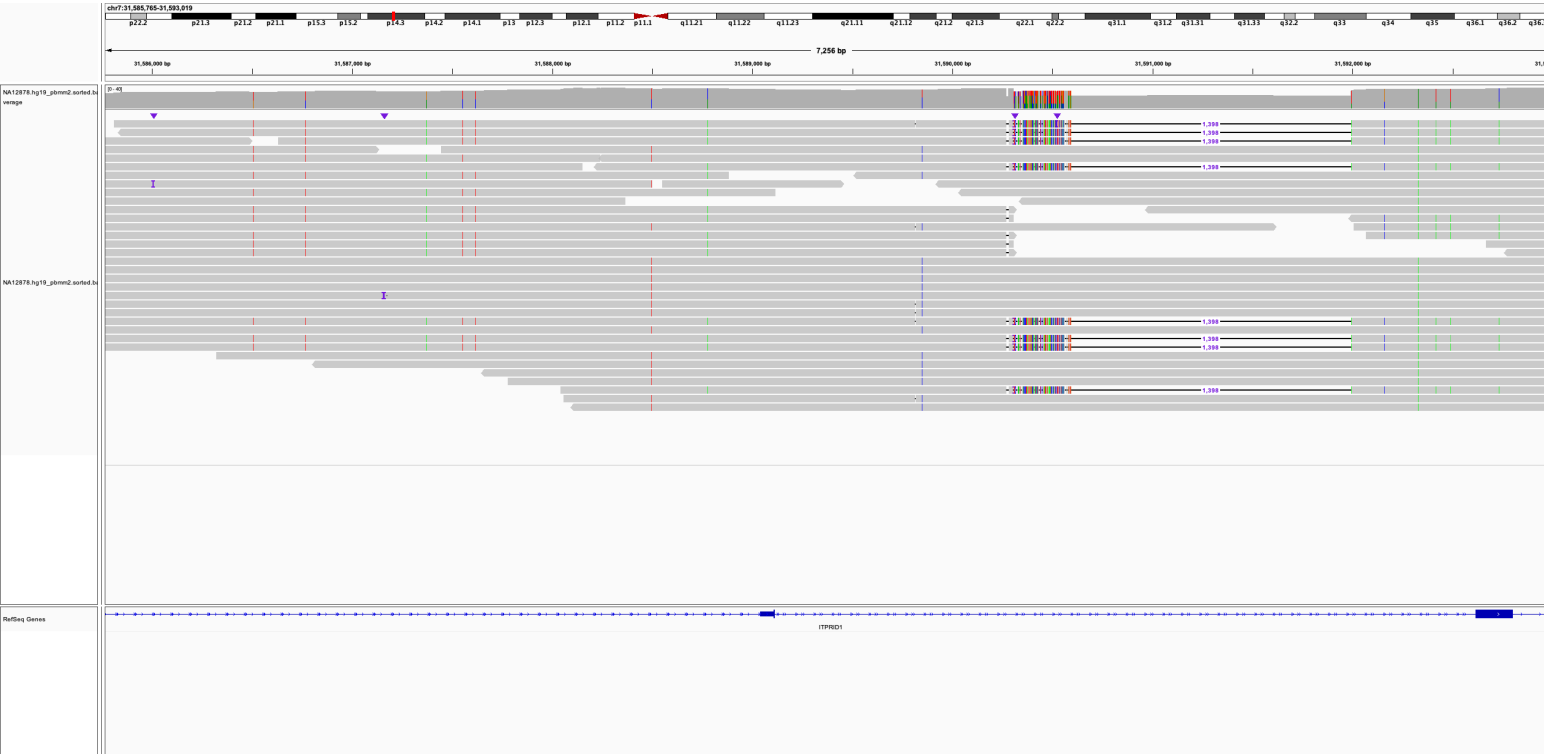
❖ chr5:147,552,866-147,554,617



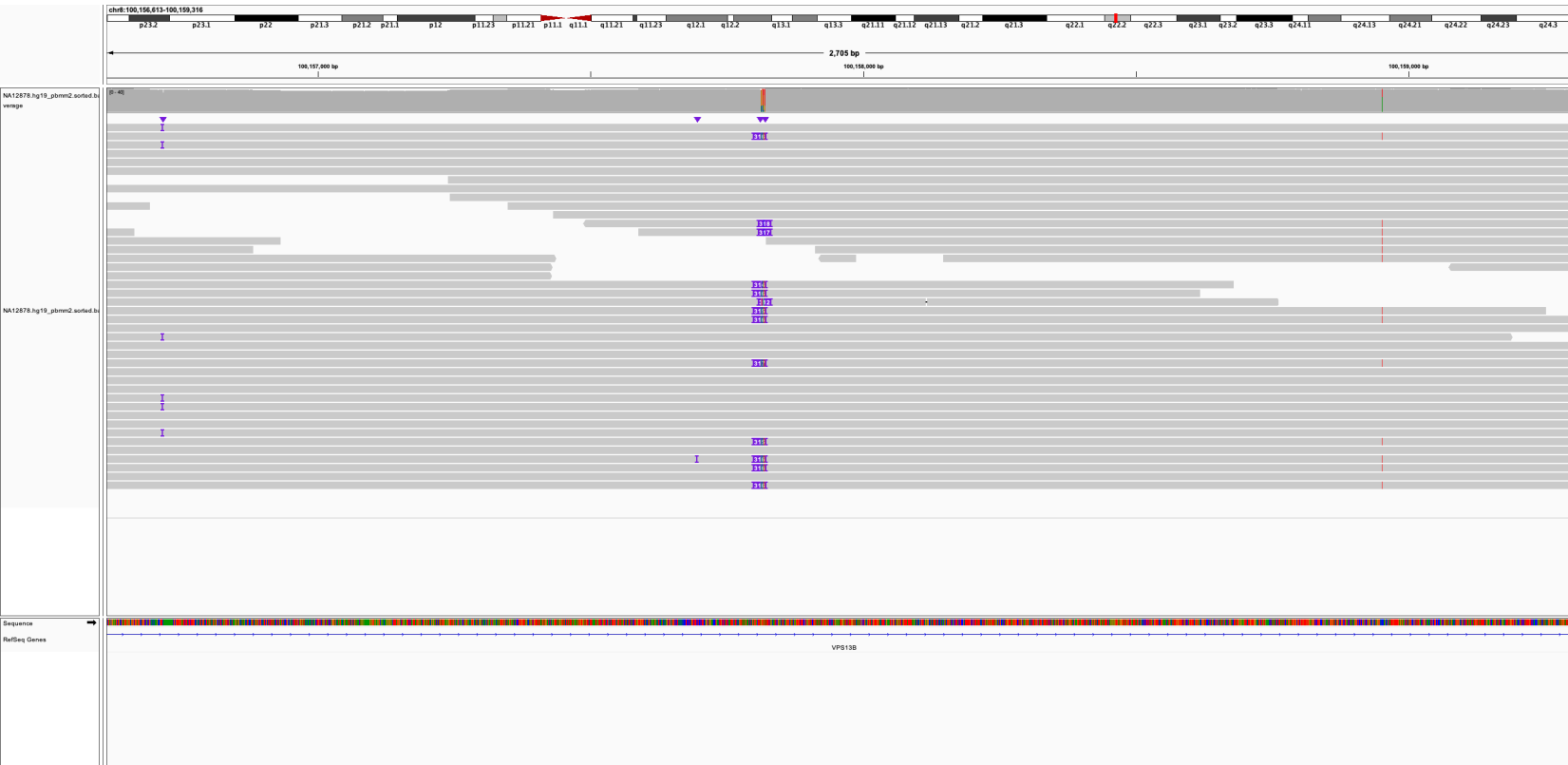
❖ chr5:169,597,462-169,598,751



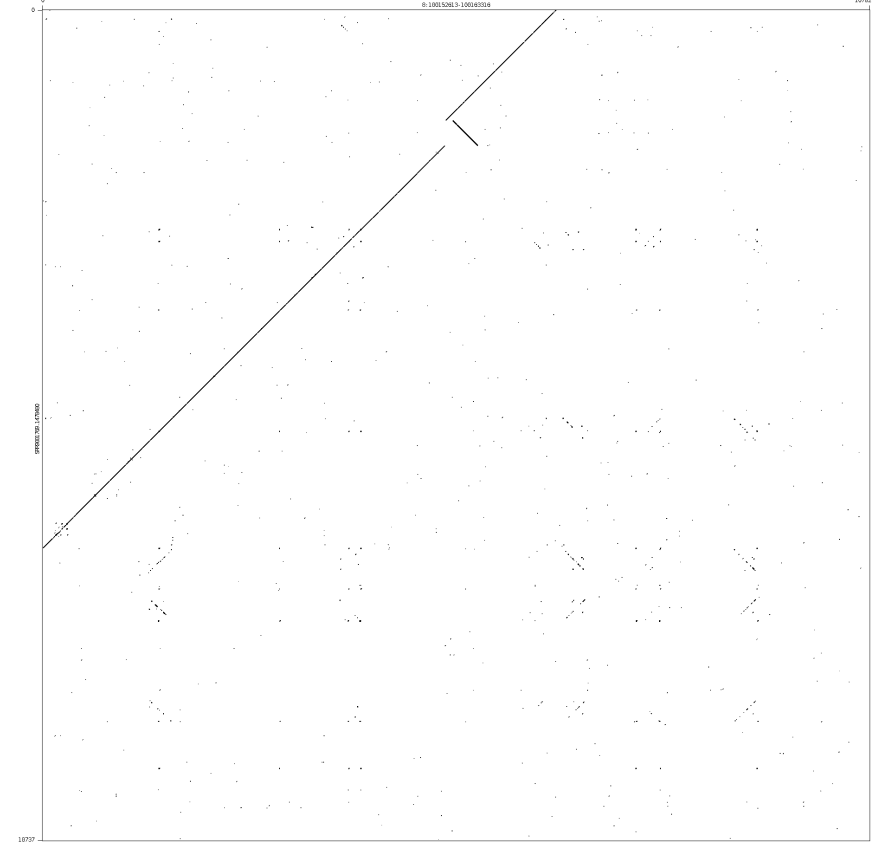
❖ chr7:31,586,765-31,592,019



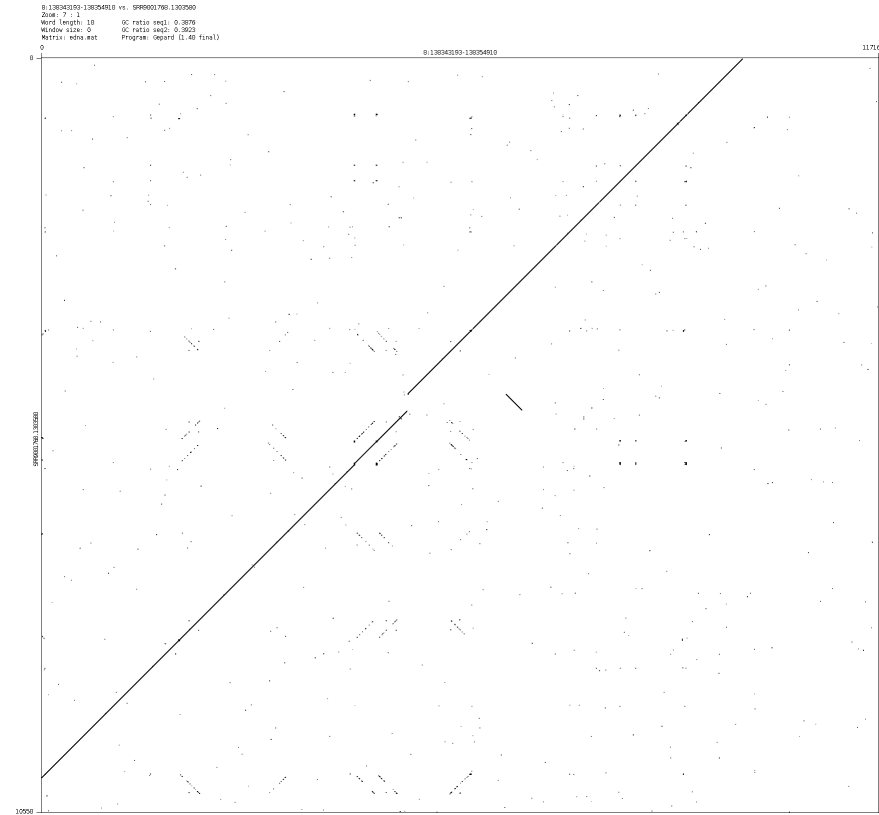
❖ chr8:100,157,613-100,158,316



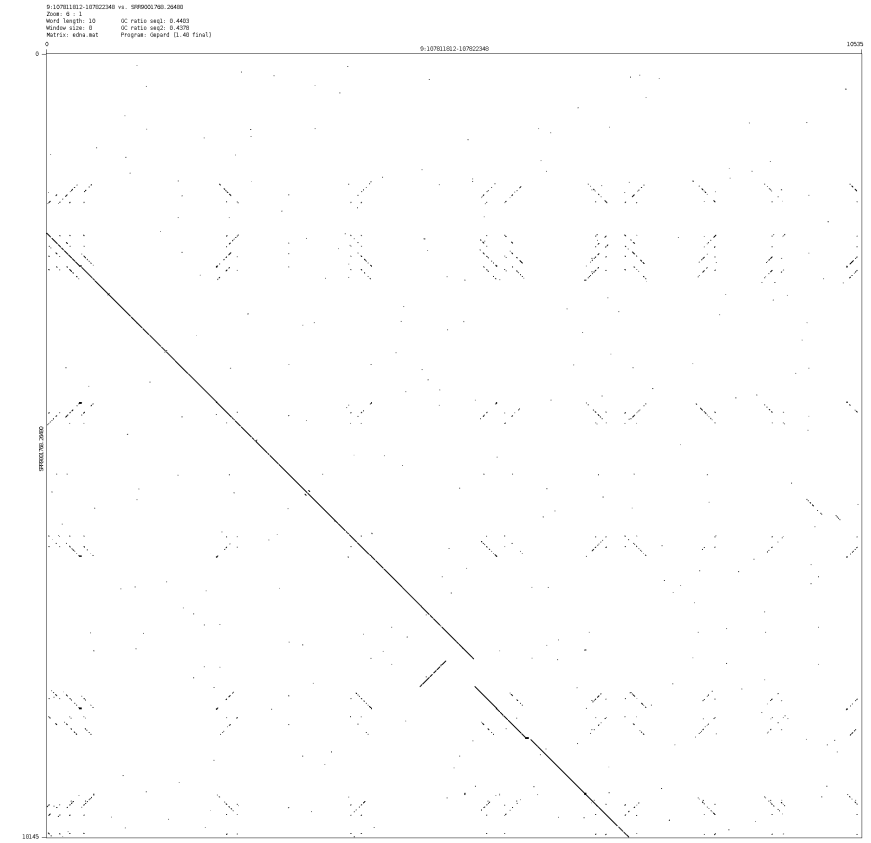
0:100152613-100152616 vs. SP95905:147940
Score: 0.1
Word Length: 30 GC ratio: 50.0
Matrix: edit-mat GC ratio: 50.0
Program: Gapped (1.40 First)



❖ chr8:138,348,193-138,349,910

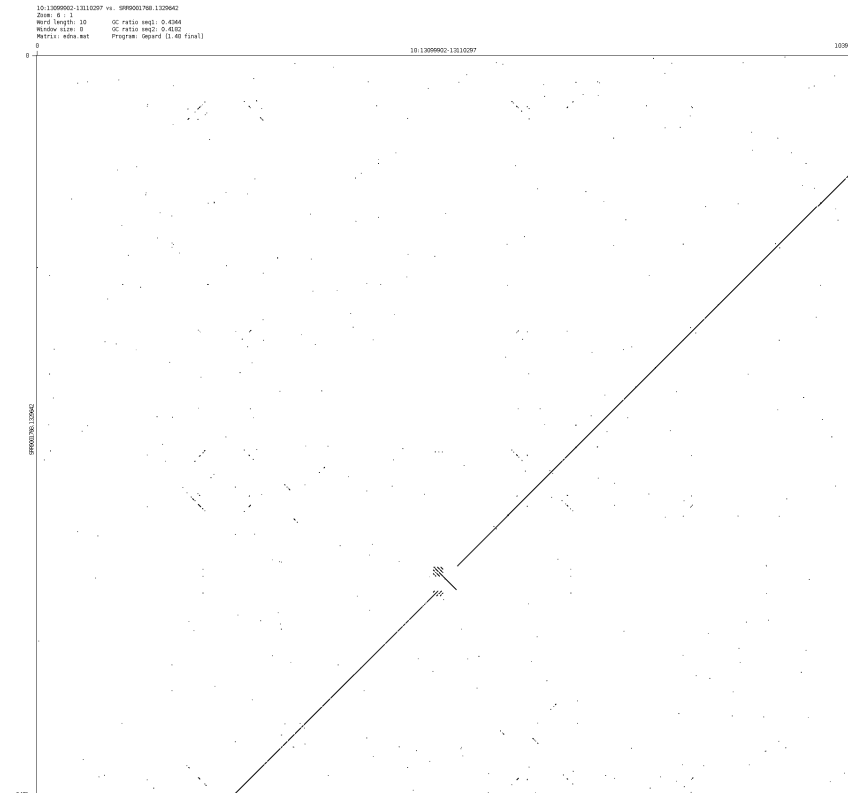


❖ chr9:107,816,812-107,817,348



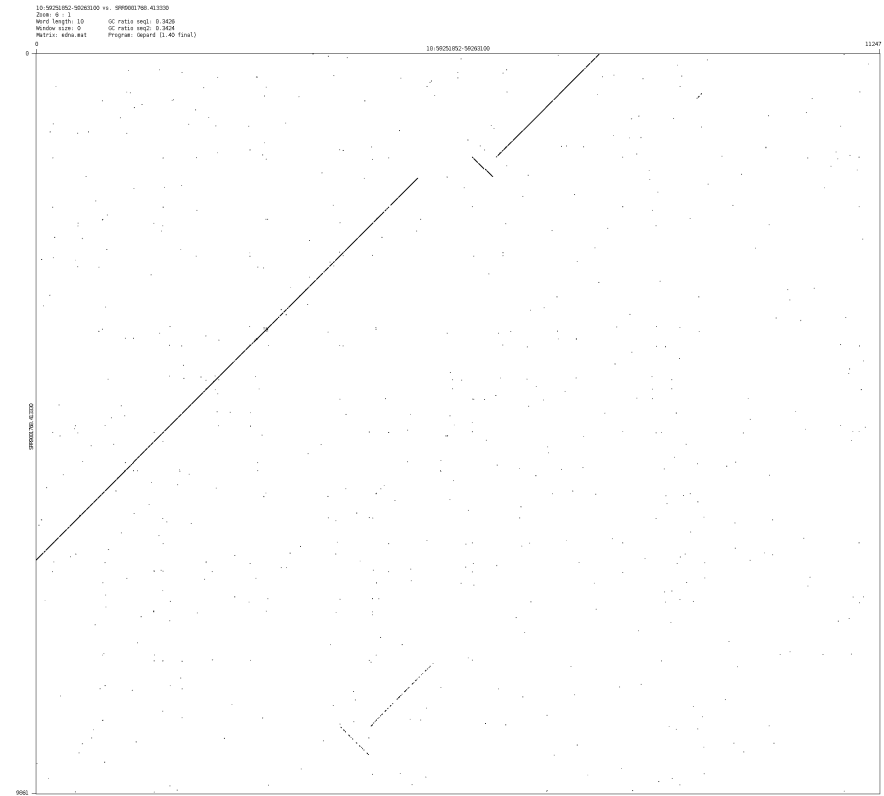
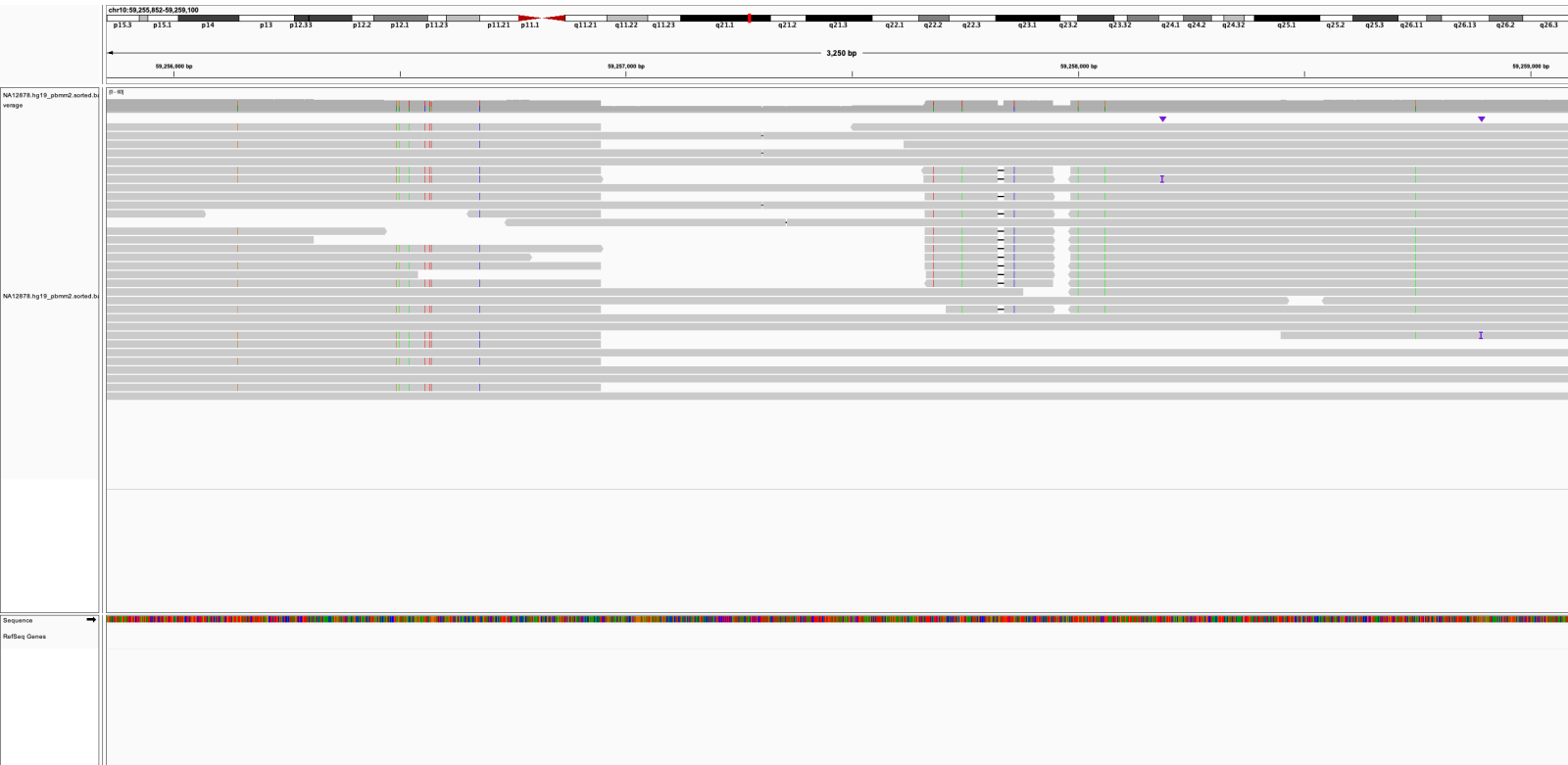


chr10:13,104,902-13,105,297

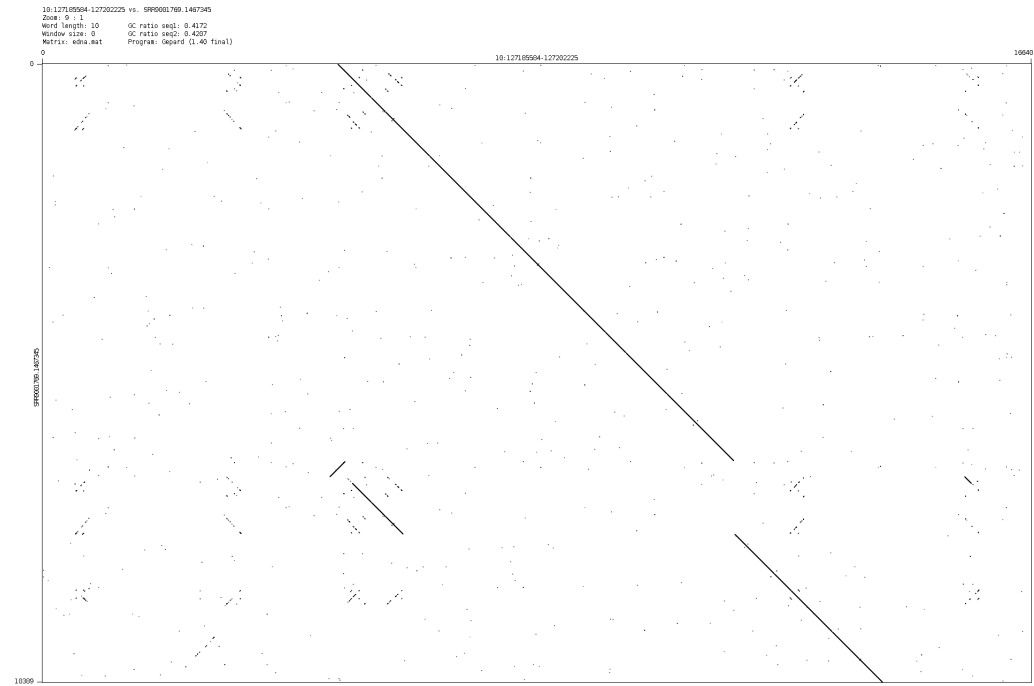
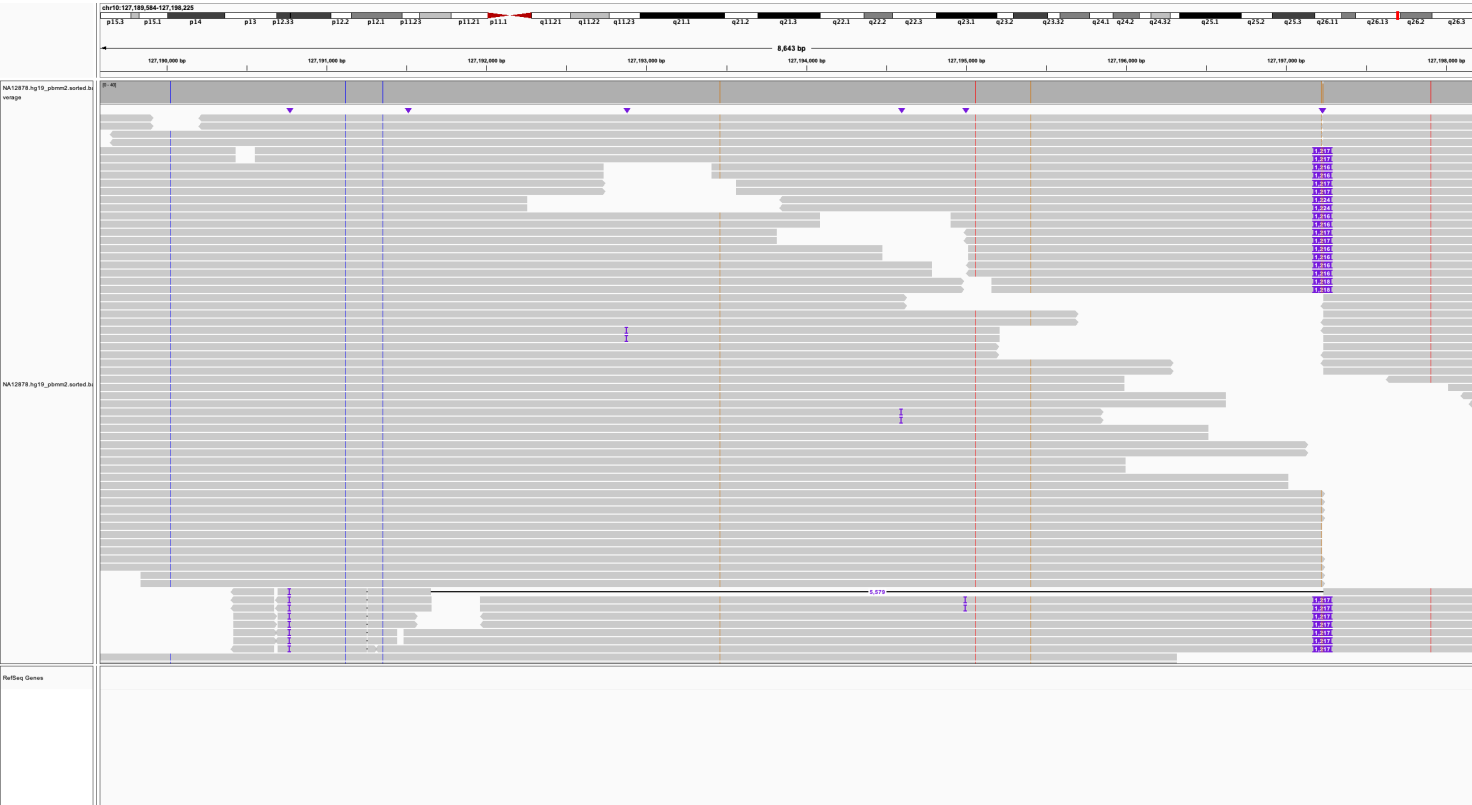




chr10:59,256,852-59,258,100

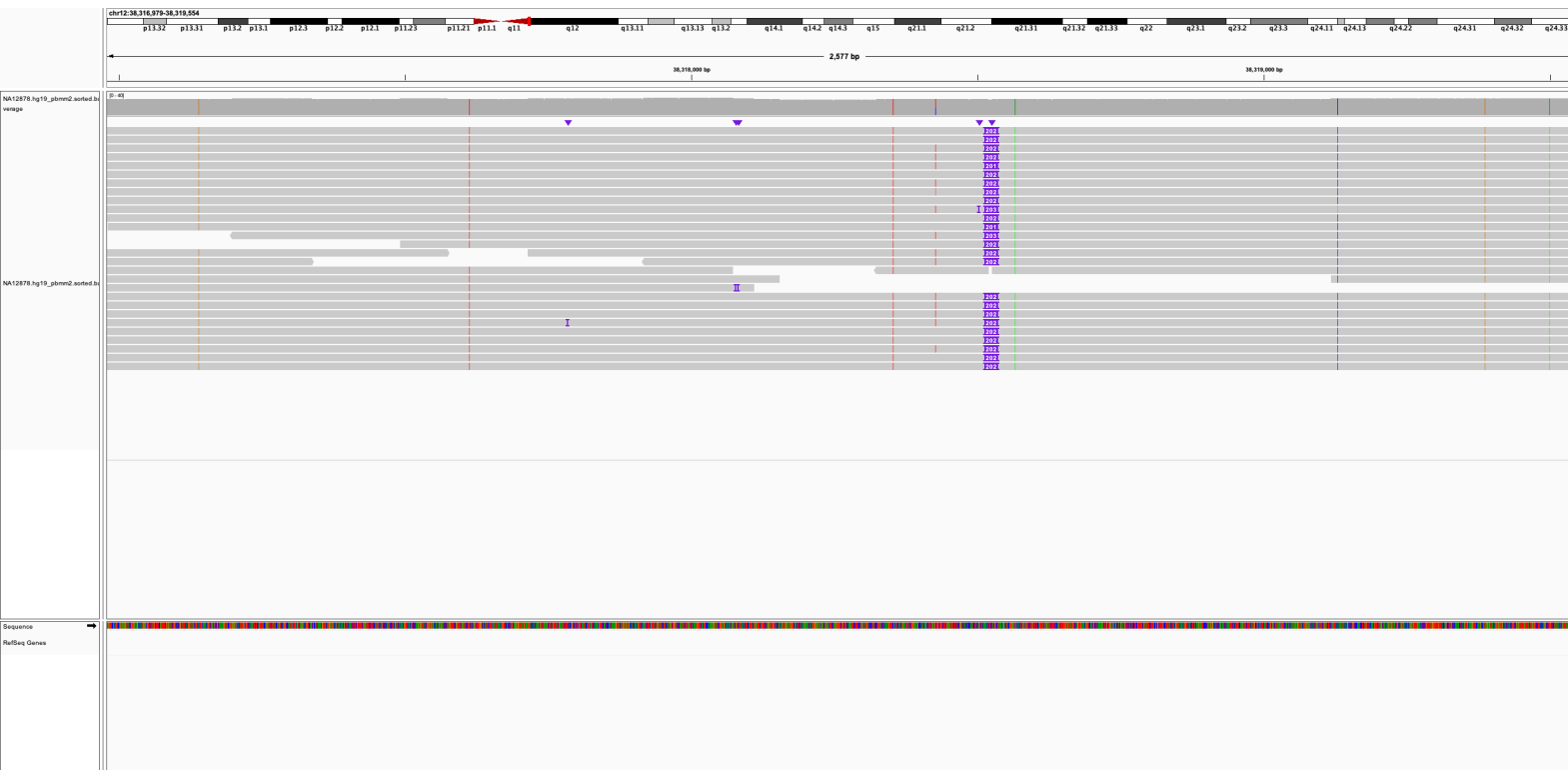


❖ chr10:127,190,584-127,197,225





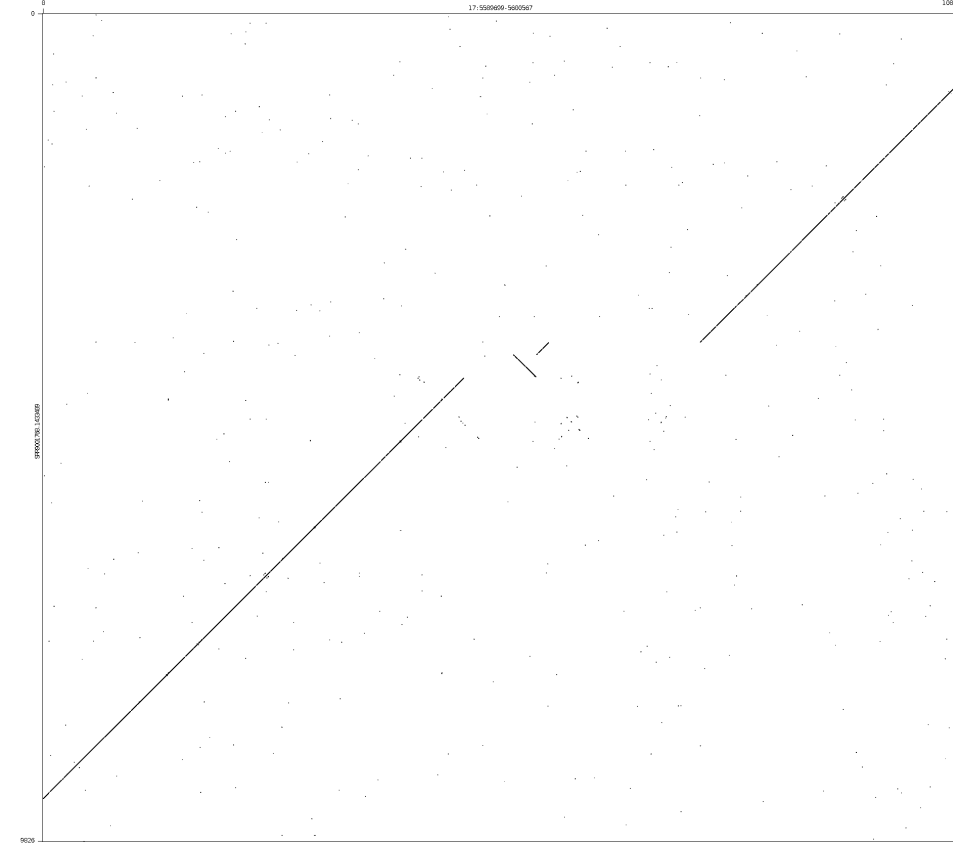
chr12:38,317,979-38,318,554



❖ chr17:5,594,699-5,595,567



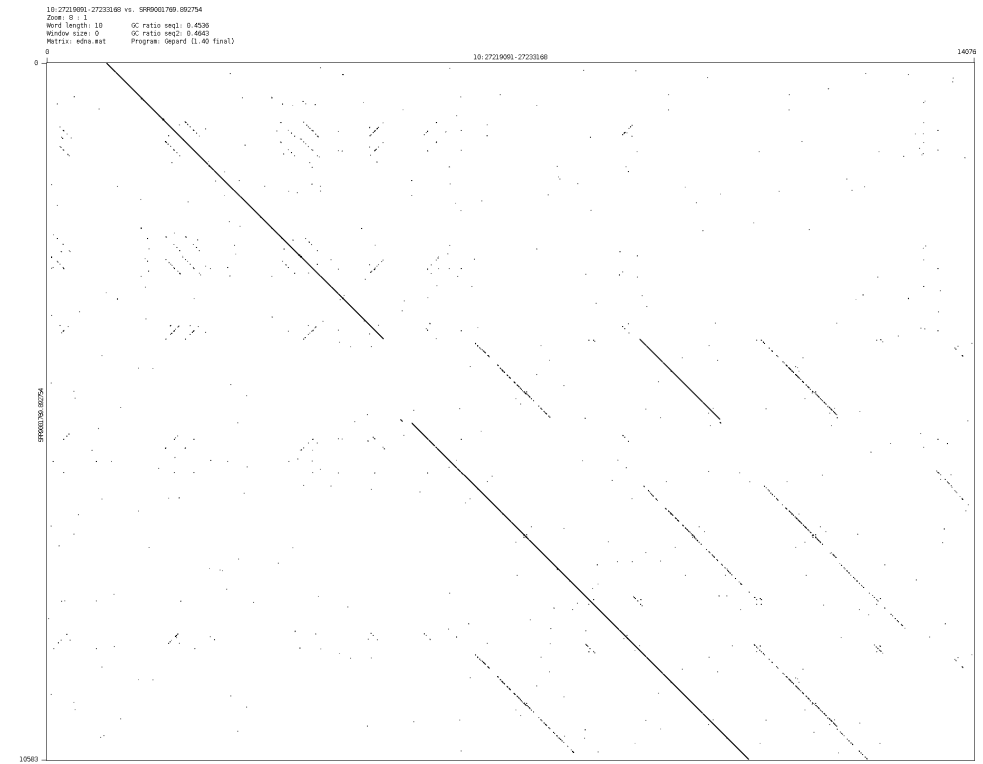
17:5594699-5600567 vs. SRR000769.1433409
Score: 0.1
Word length: 10 GC ratio seq: 0.4807
Window size: 5 GC ratio seq: 0.4803
Matrix: edna.mat Program: gepard [L:48 Final]



Simple duplication

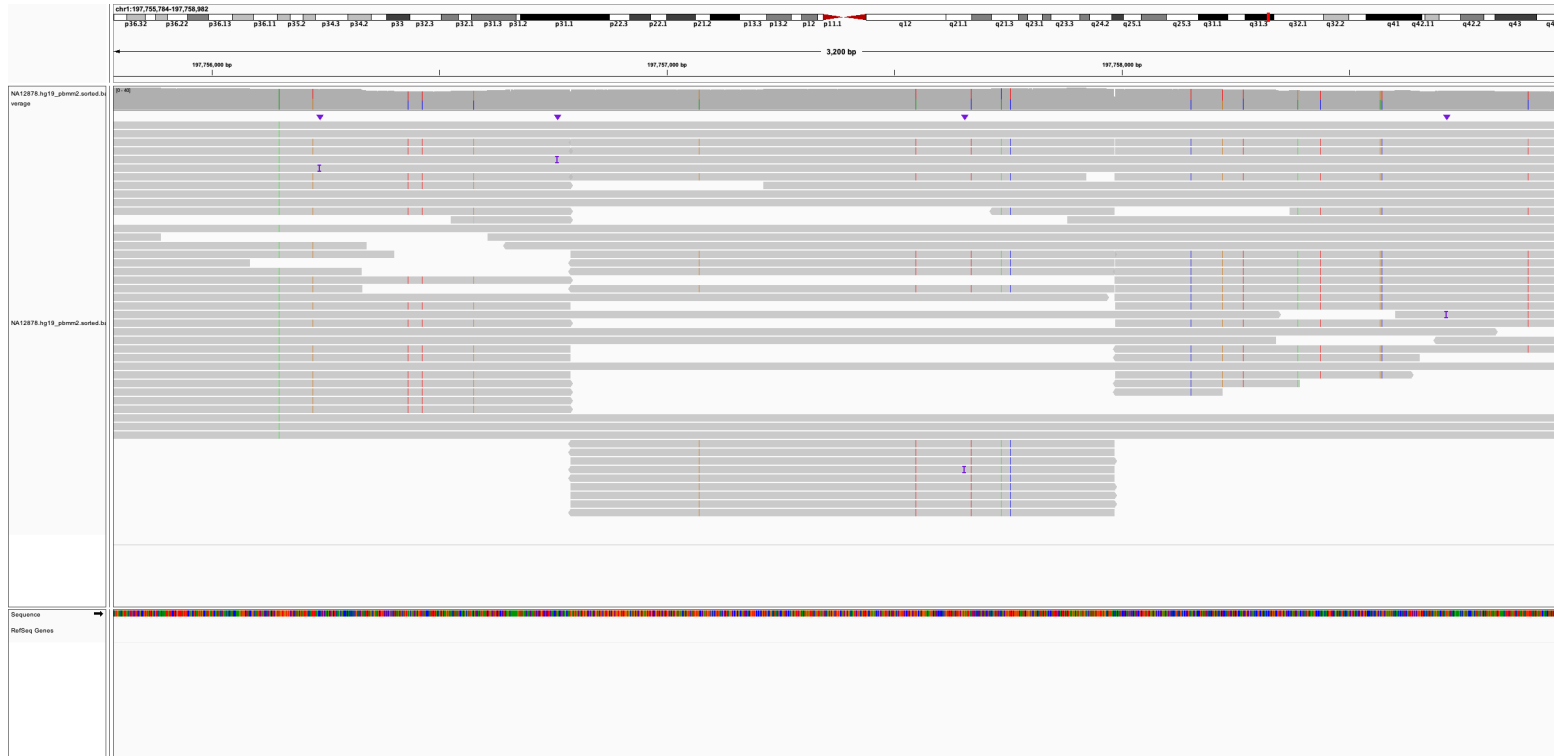


chr10:27,224,091-27,228,168

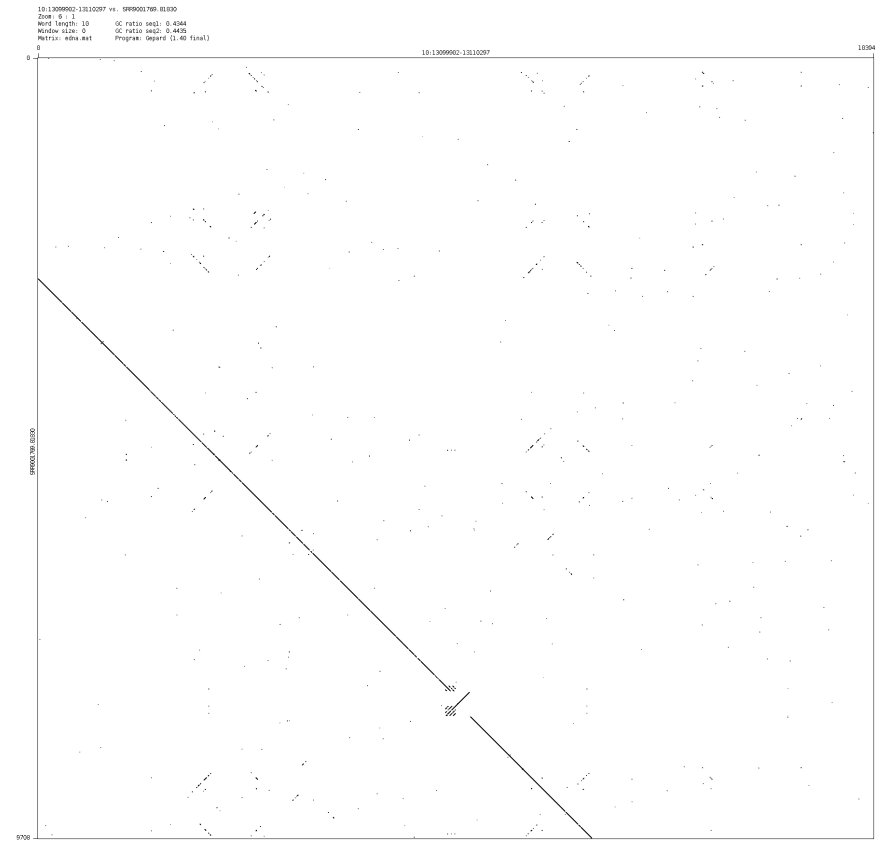
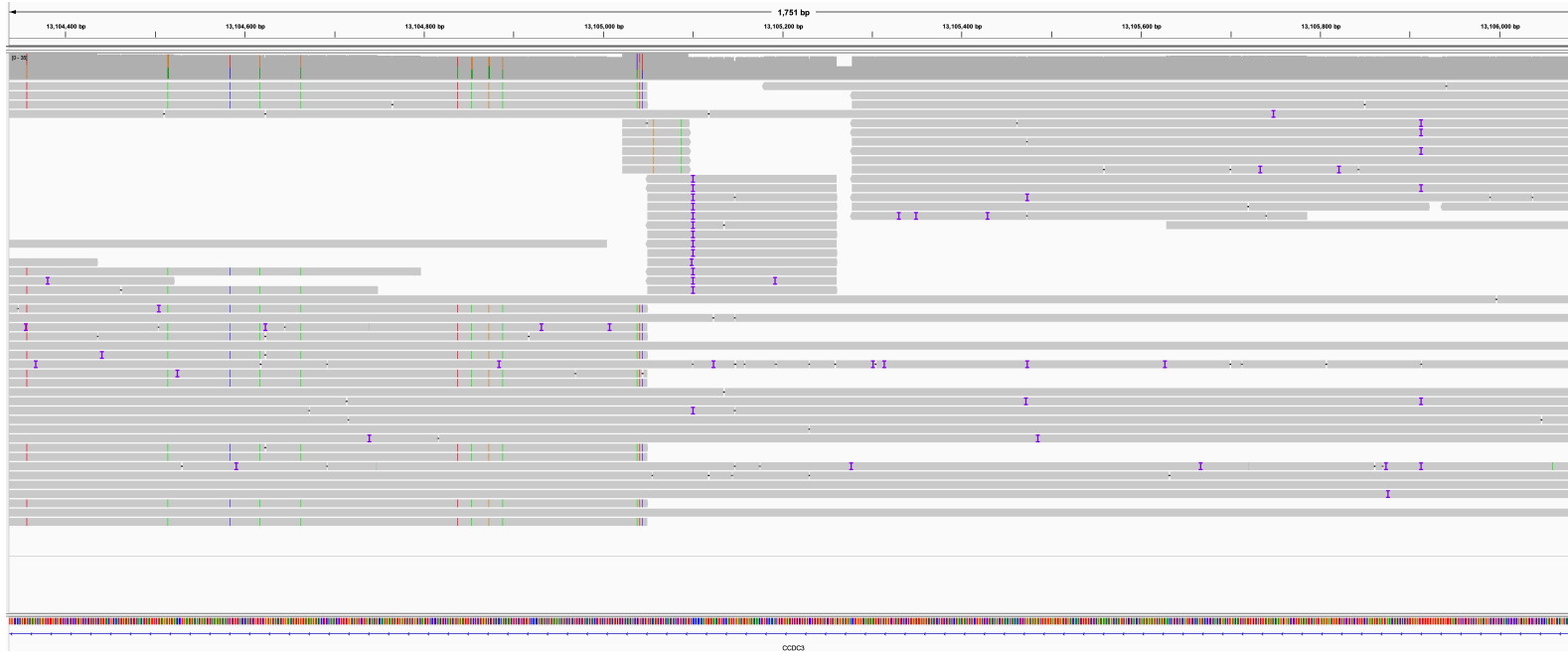


Simple inversion

❖ chr1:197,756,784-197,757,982

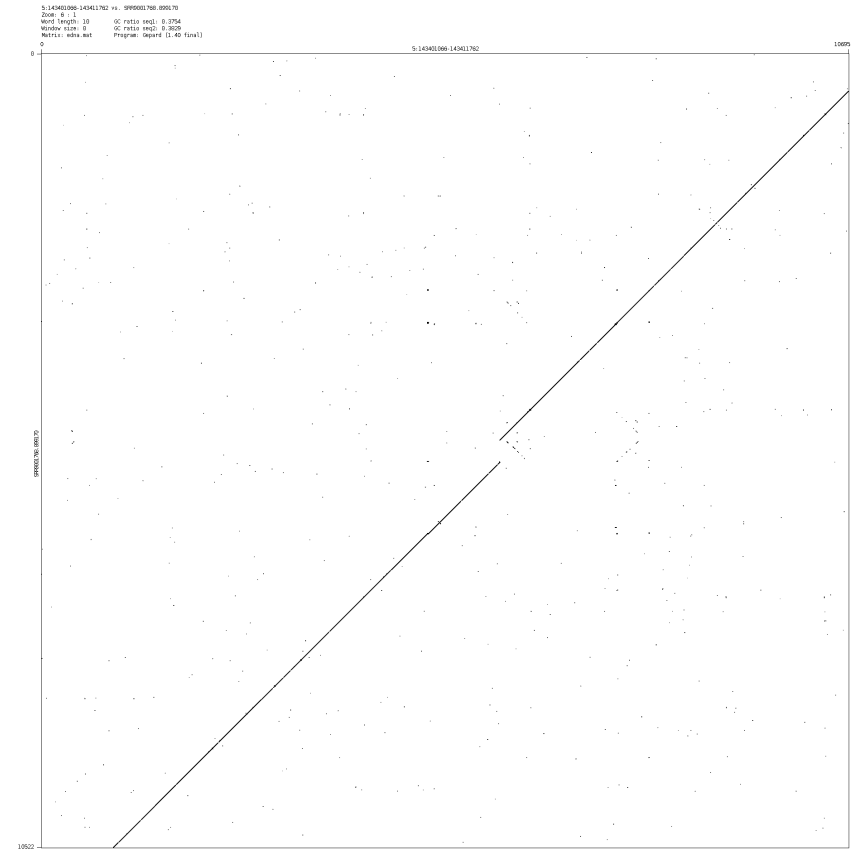
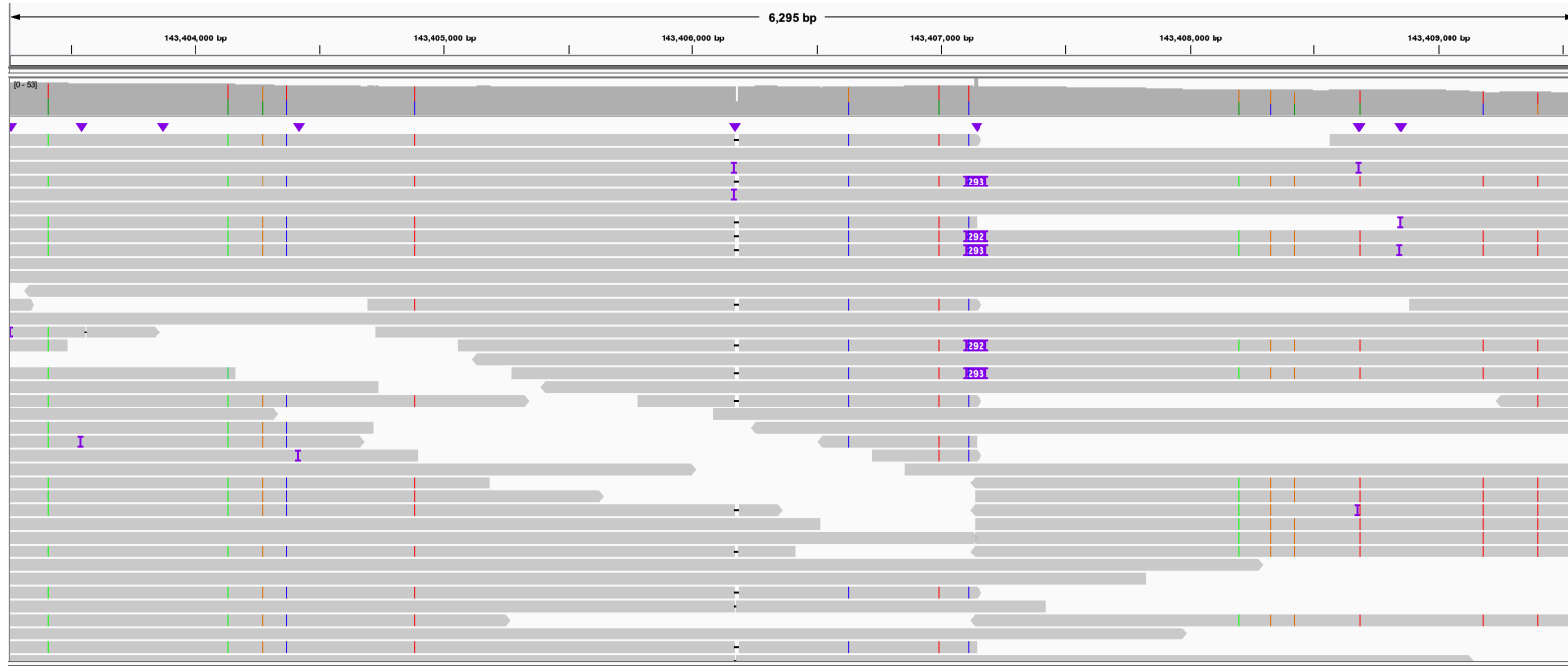


❖ chr10:13,104,902-13,105,297

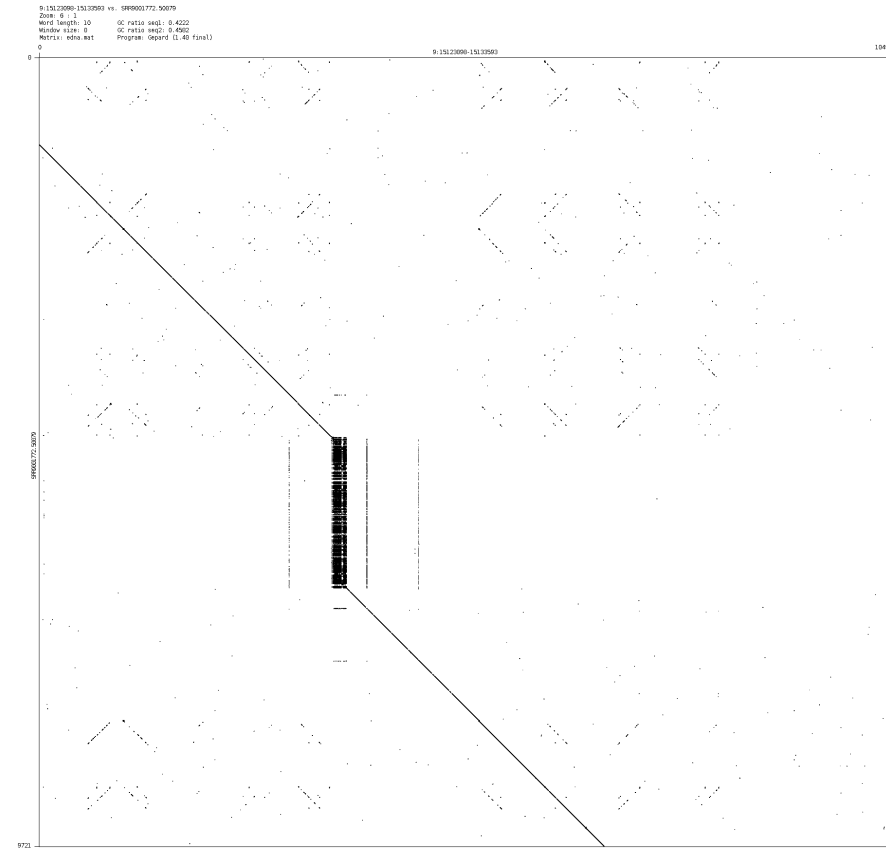
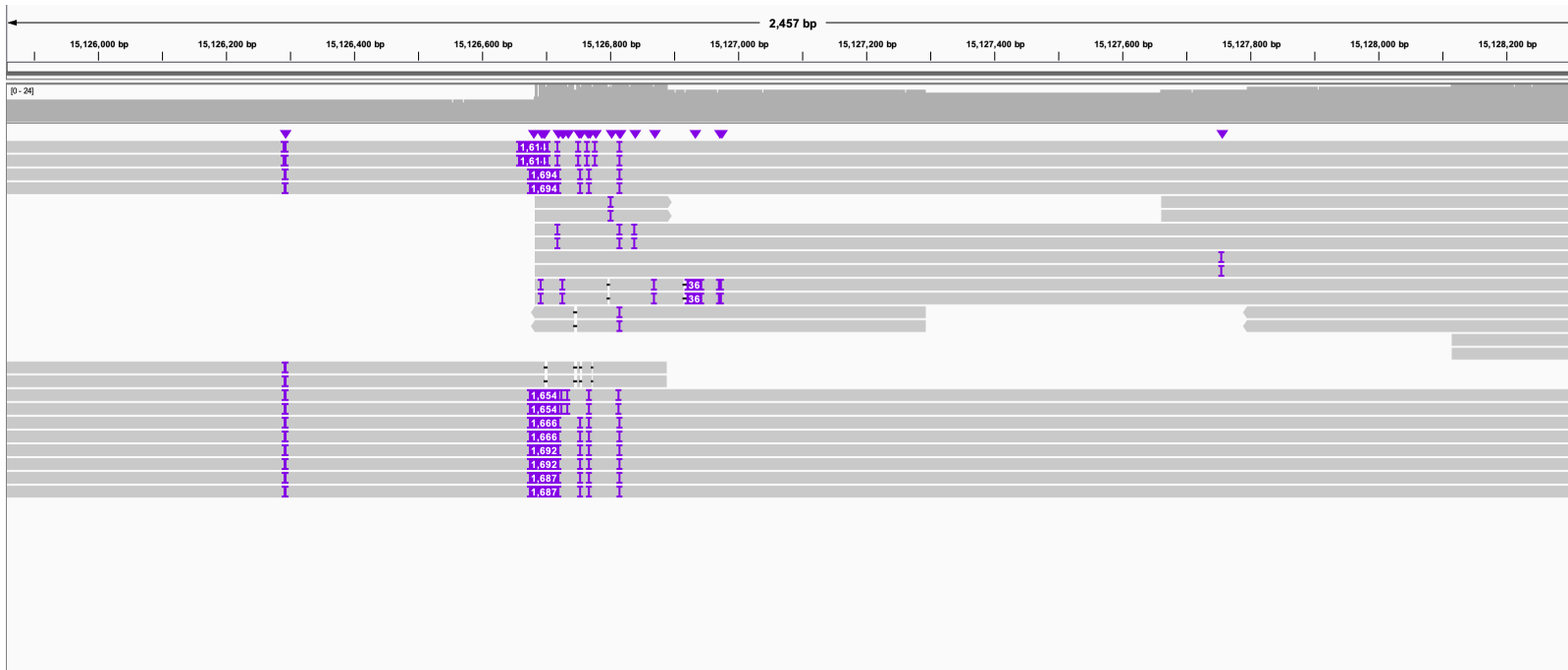


Simple insertion

❖ chr5:143,406,066-143,406,762

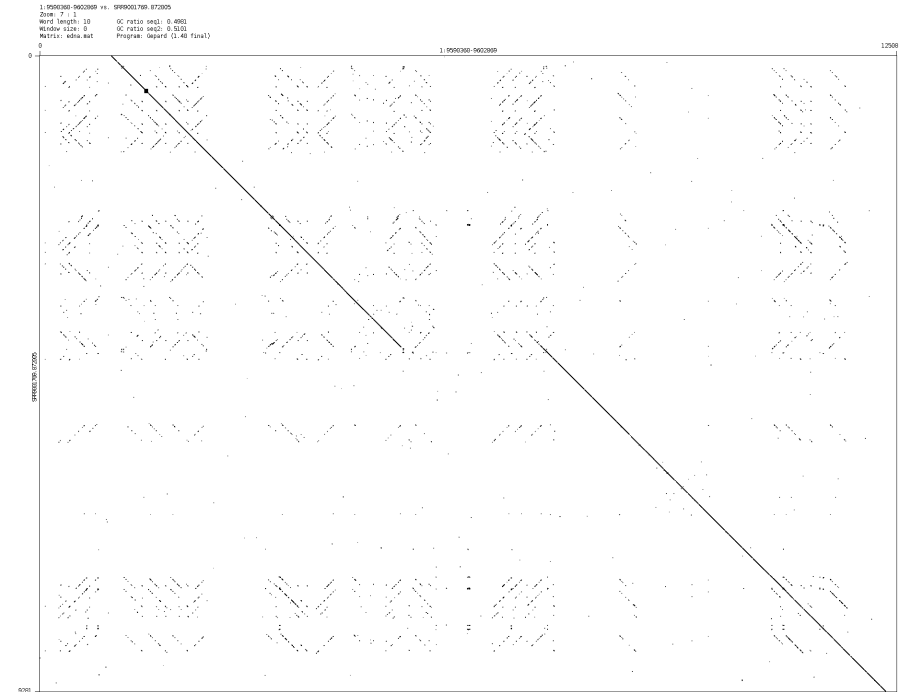
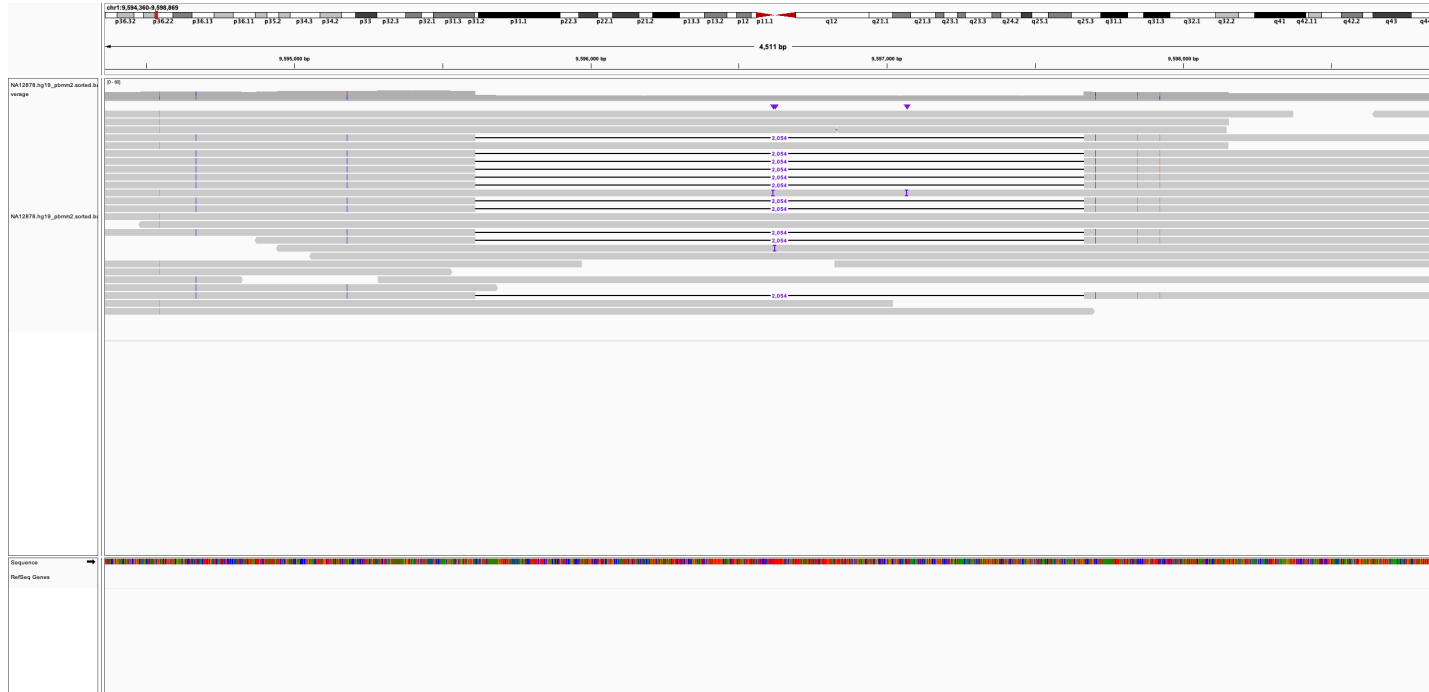


❖ chr9:15,128,098-15,128,593

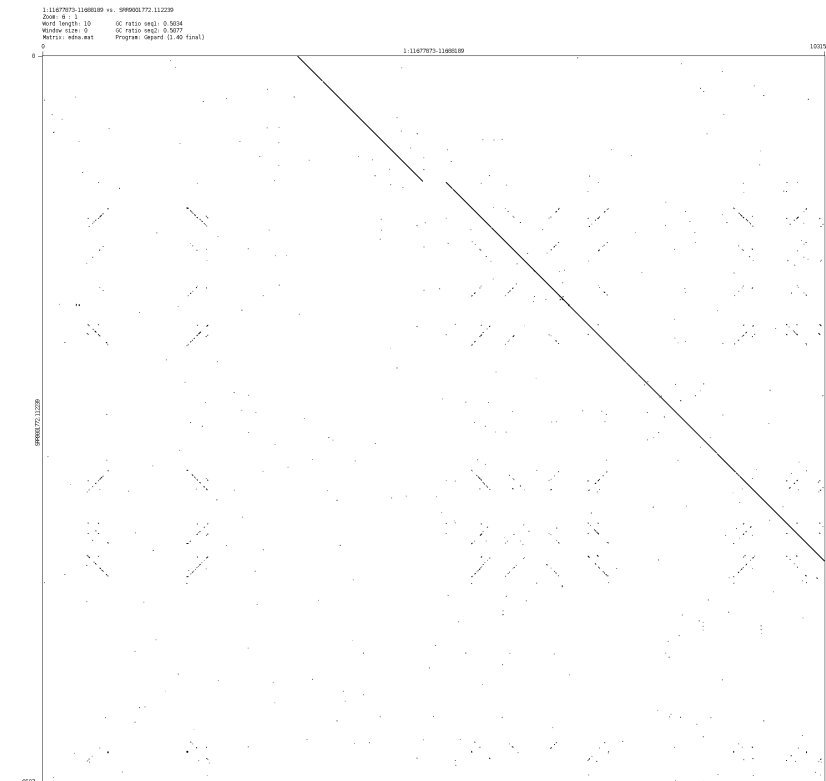
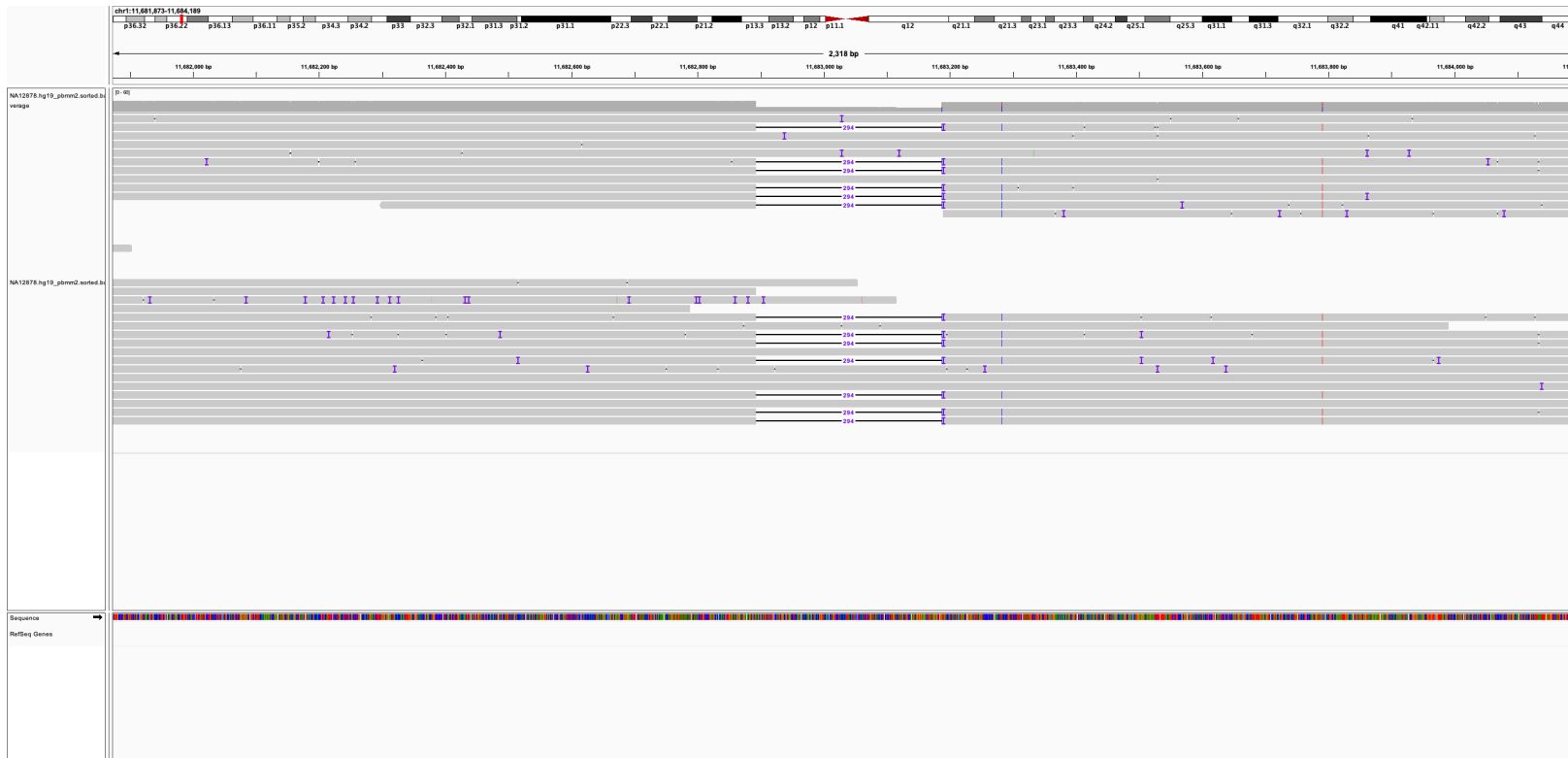


Simple deletion

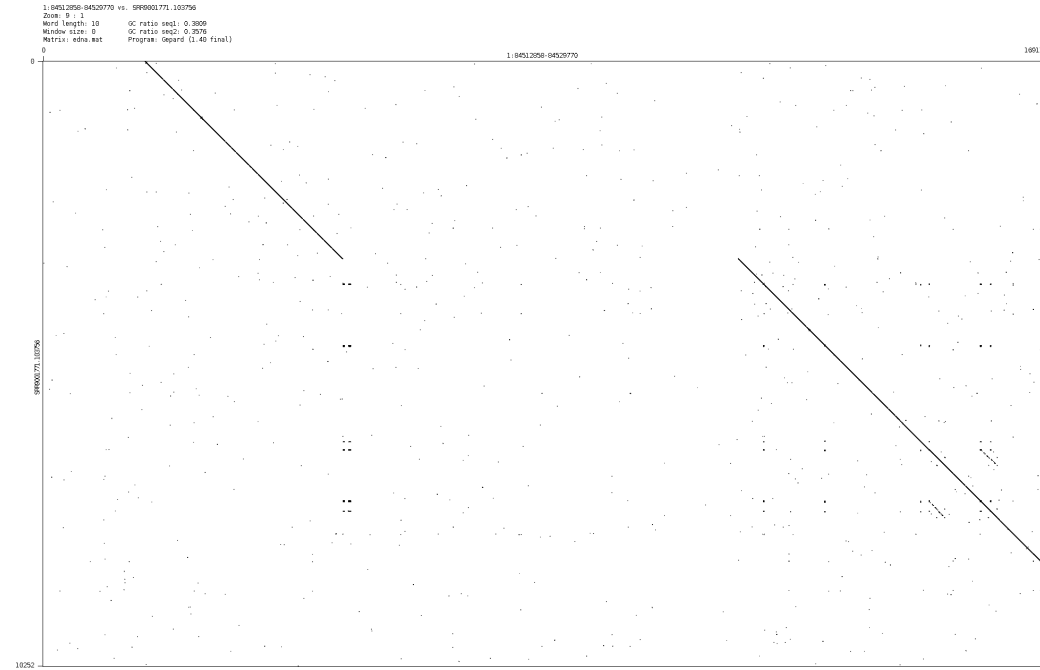
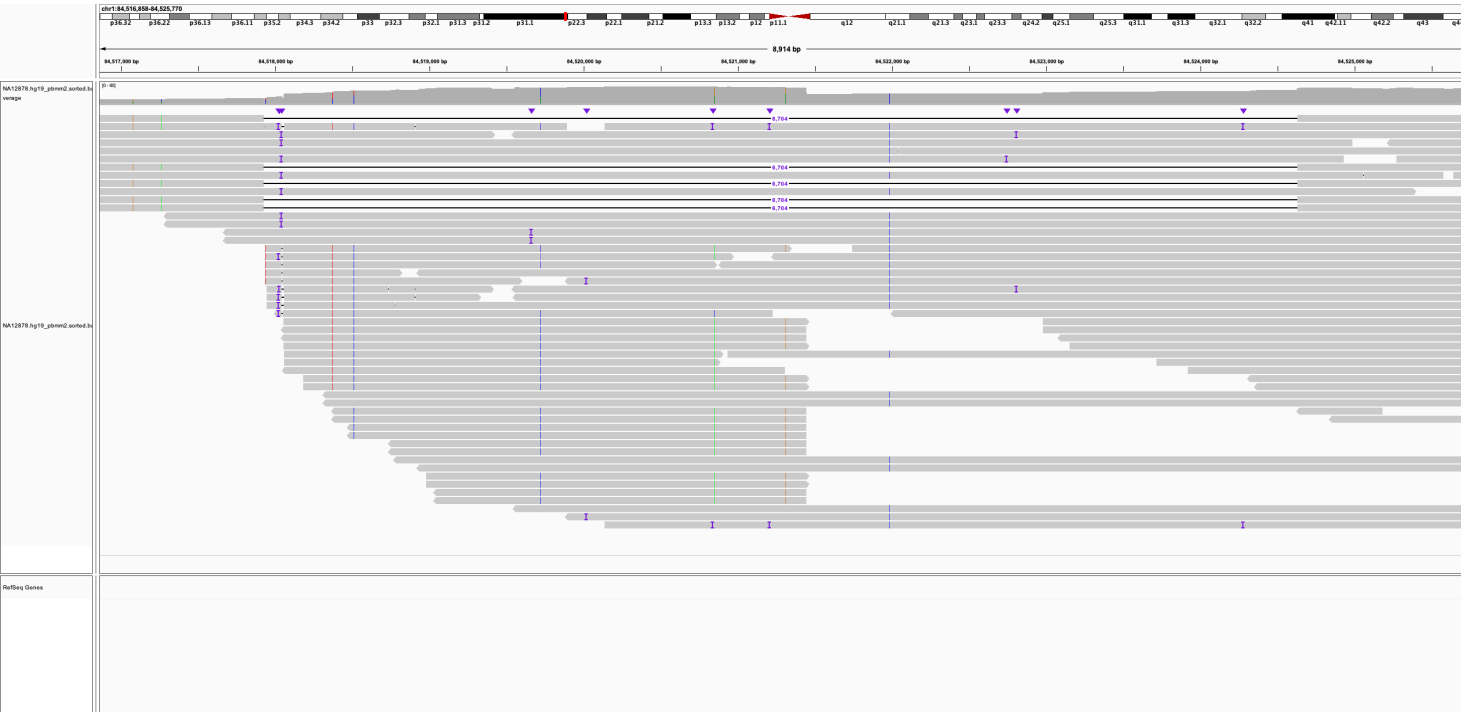
❖ chr1:9,595,360-9,597,869



❖ chr1:11,682,873-11,683,189



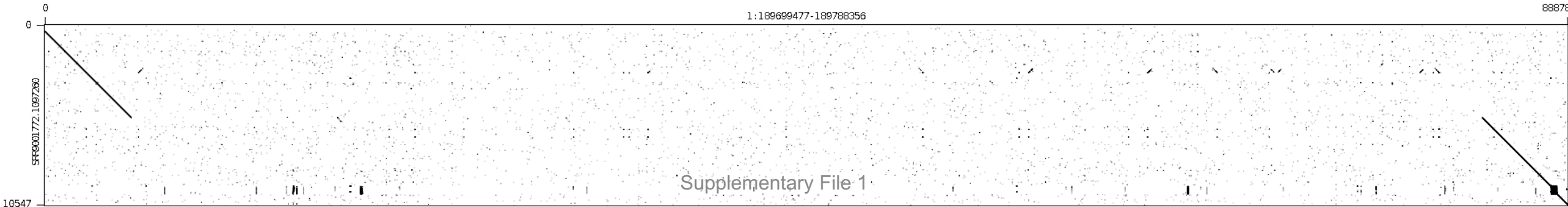
❖ chr1:84,517,858-84,524,770



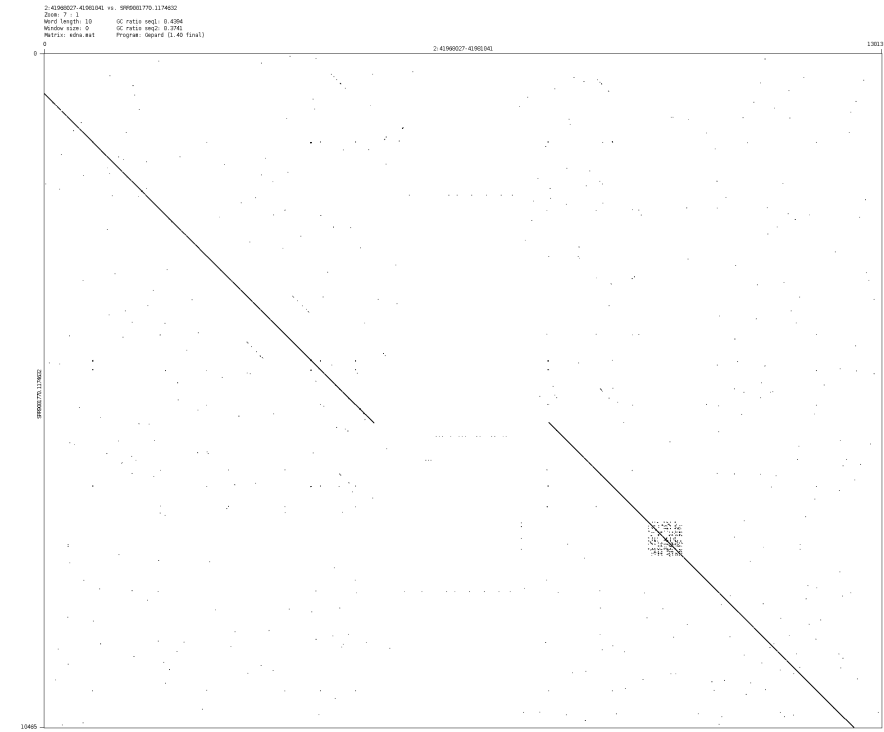
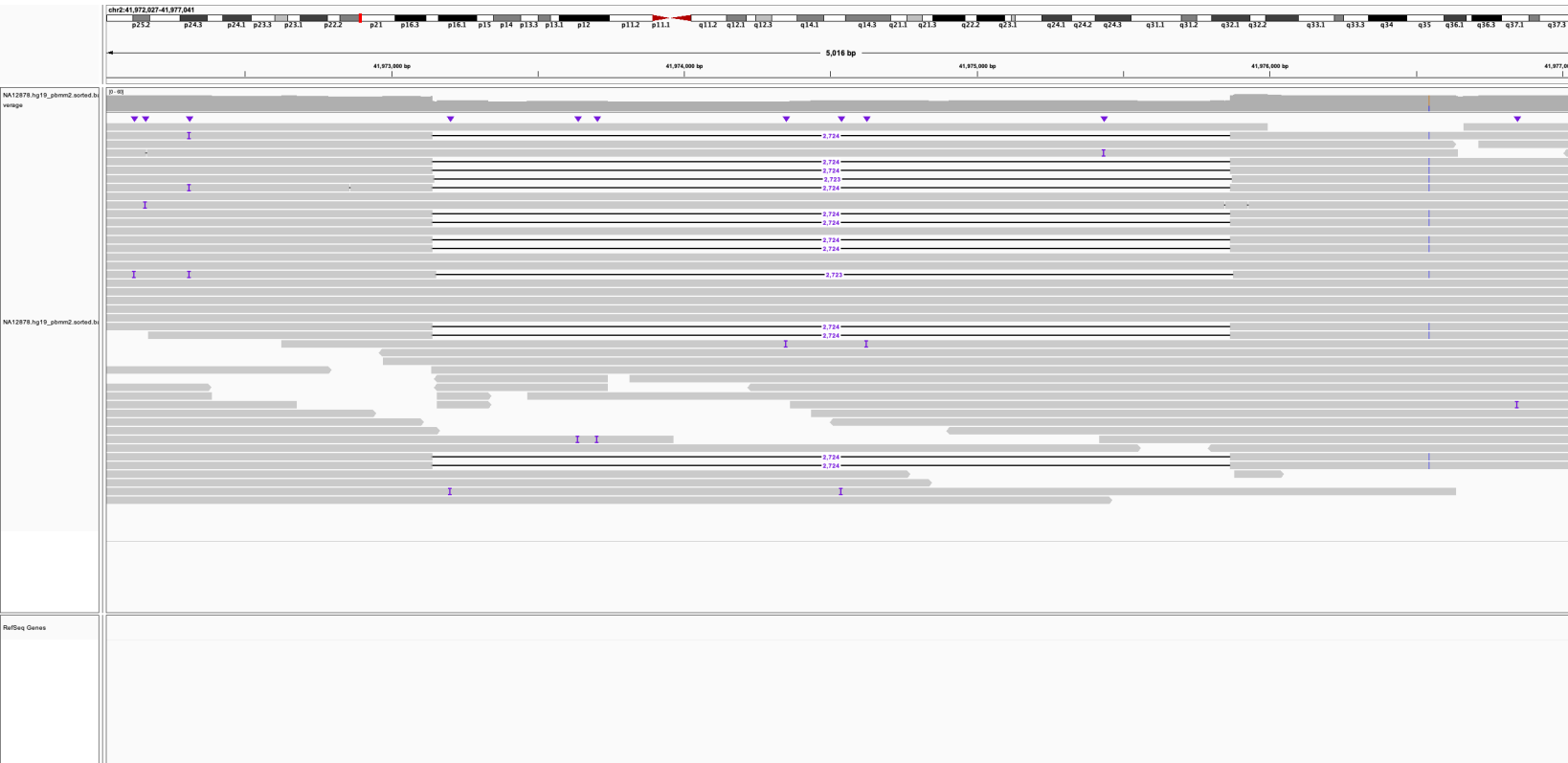
❖ chr1:189,704,477-189,783,356



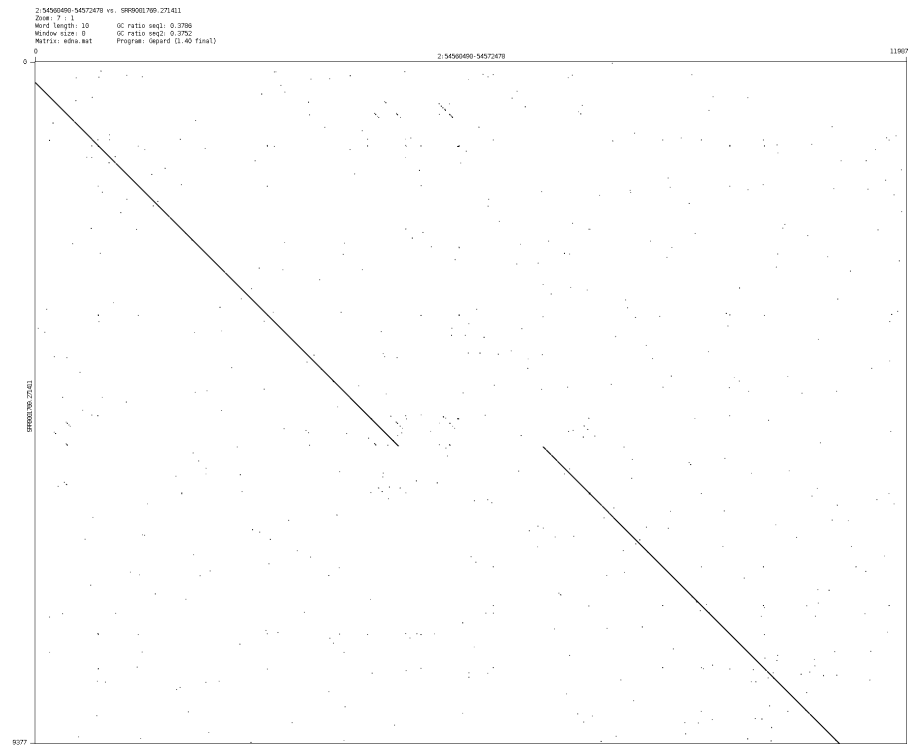
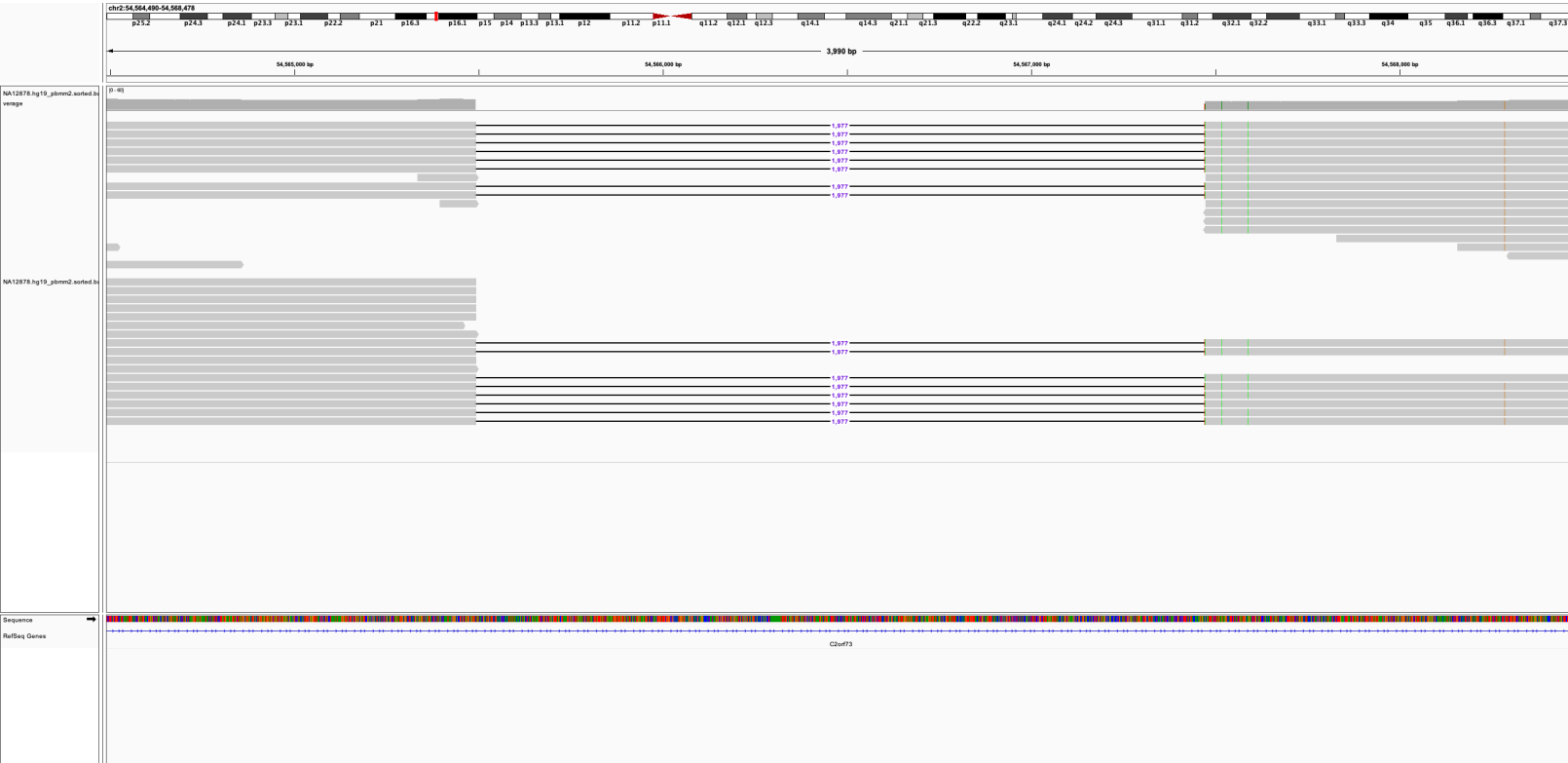
1:189699477-189788356 vs. SRR001772.1097260
Zoom: 47 : 1
Word length: 10 GC ratio seq1: 0.3393
Window size: 0 GC ratio seq2: 0.3218
Matrix: edna.mat Program: Gepard (1.40 final)



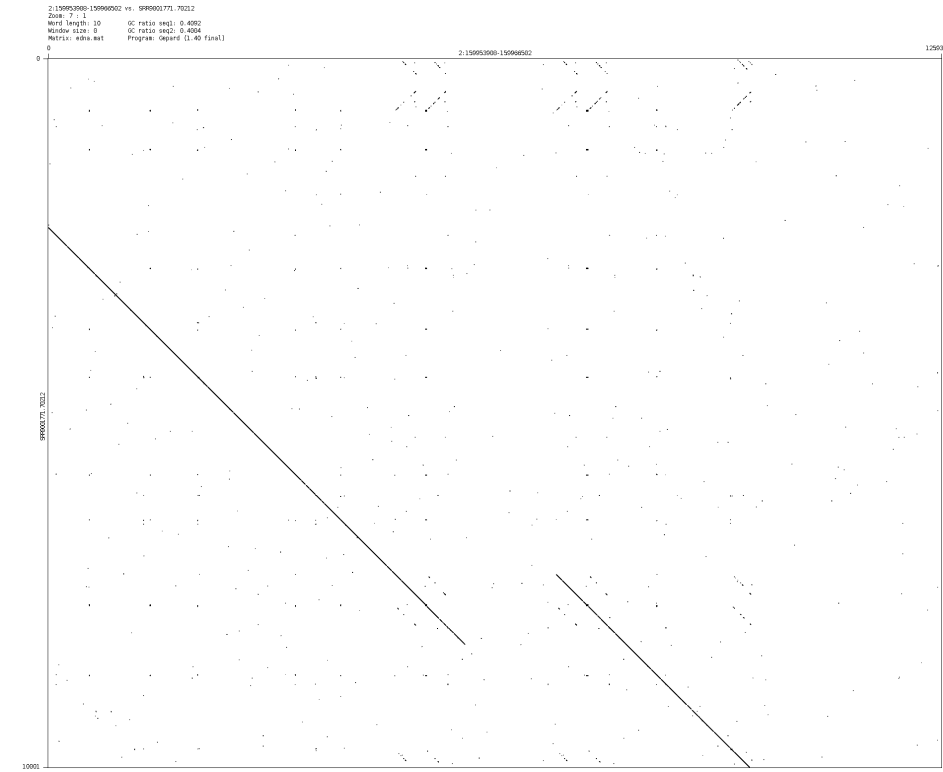
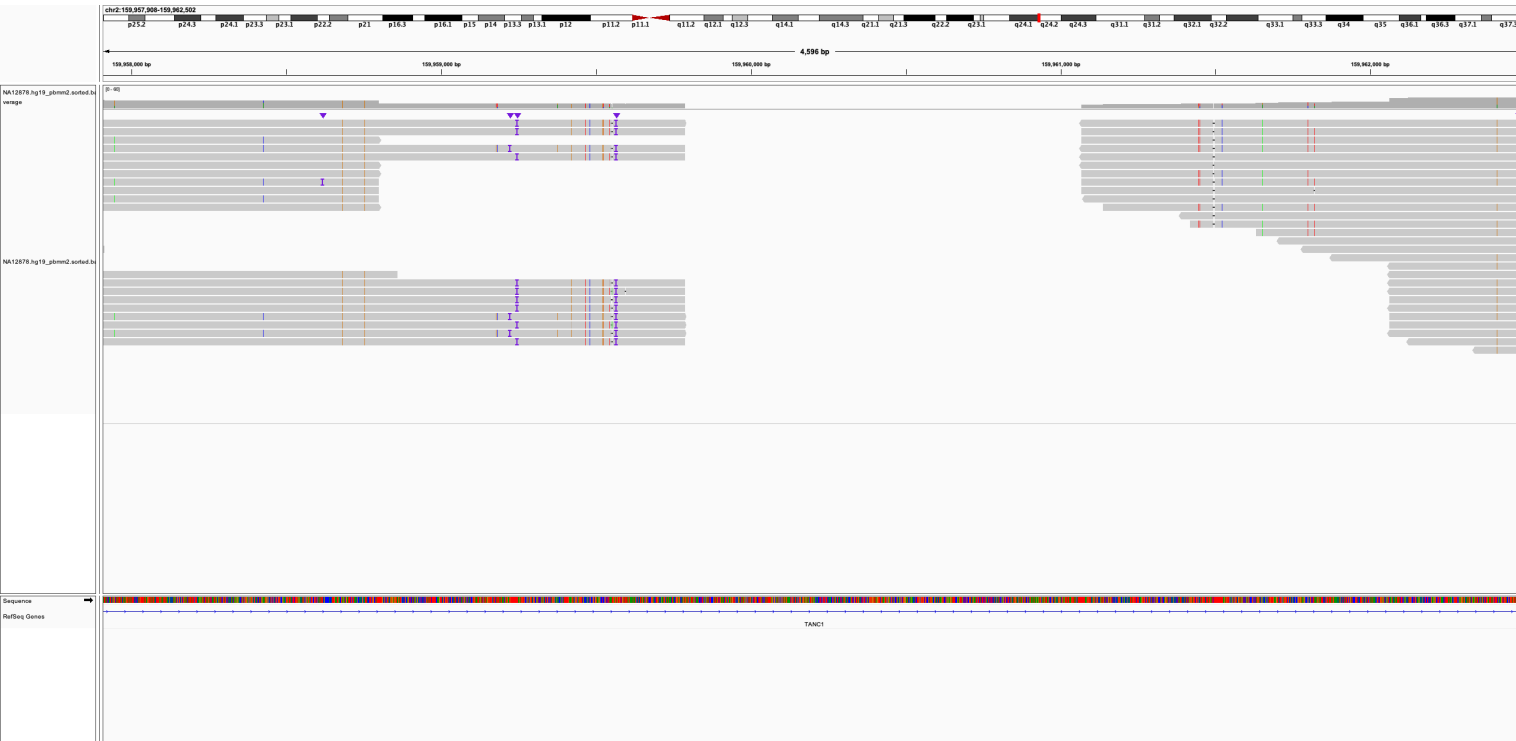
❖ chr2:41,973,027-41,976,041



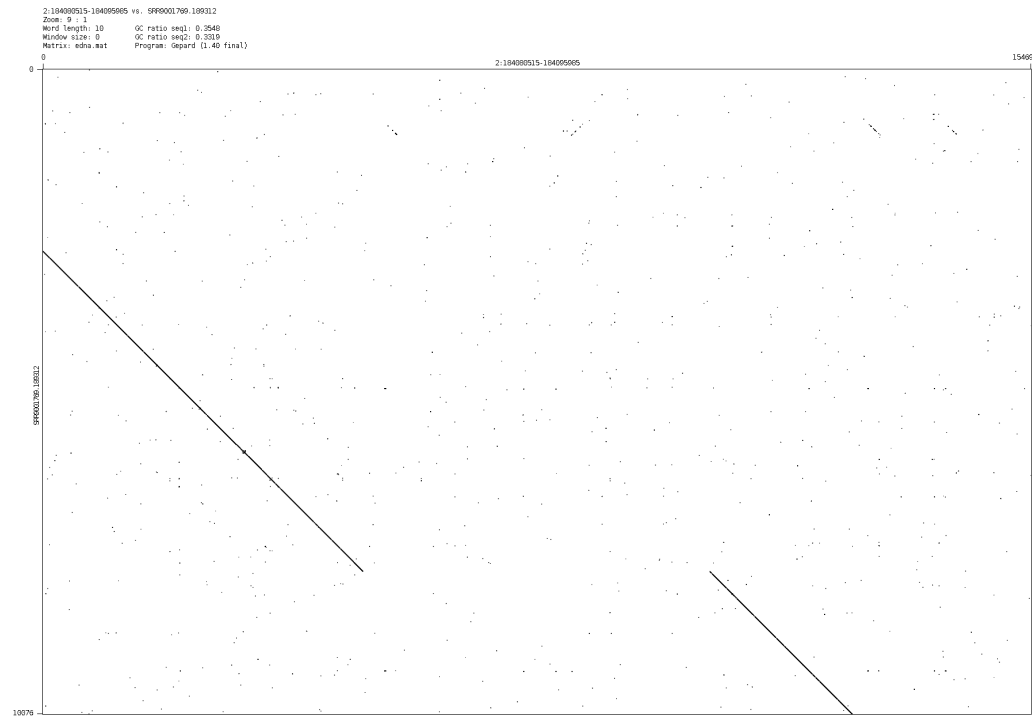
❖ chr2:54,565,490-54,567,478



❖ chr2:159,958,908-159,961,502

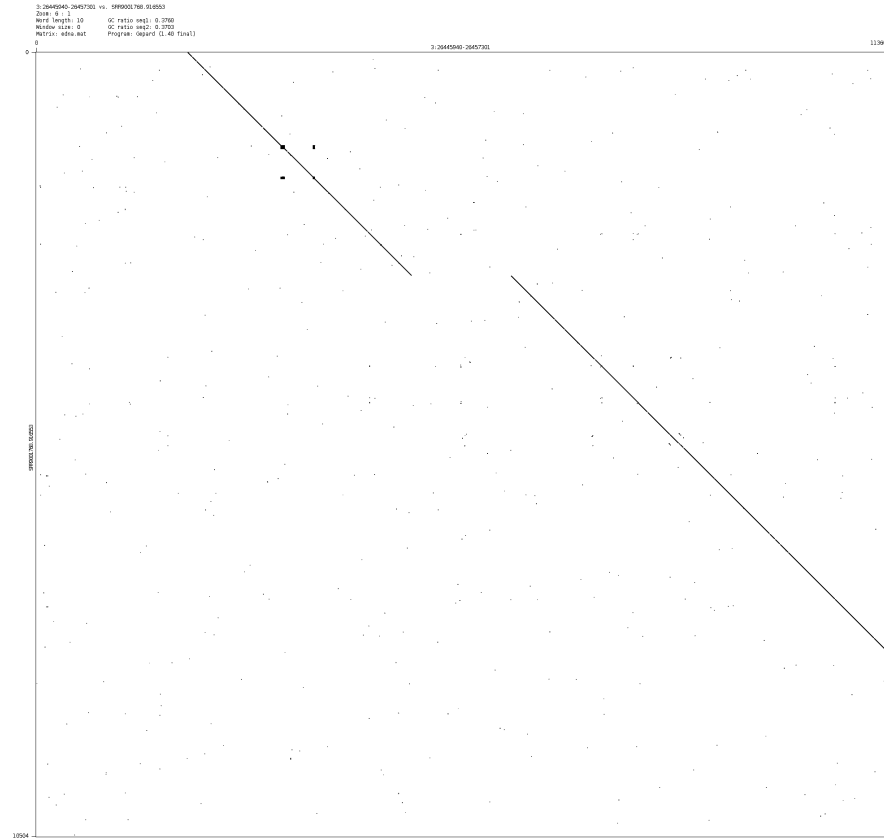
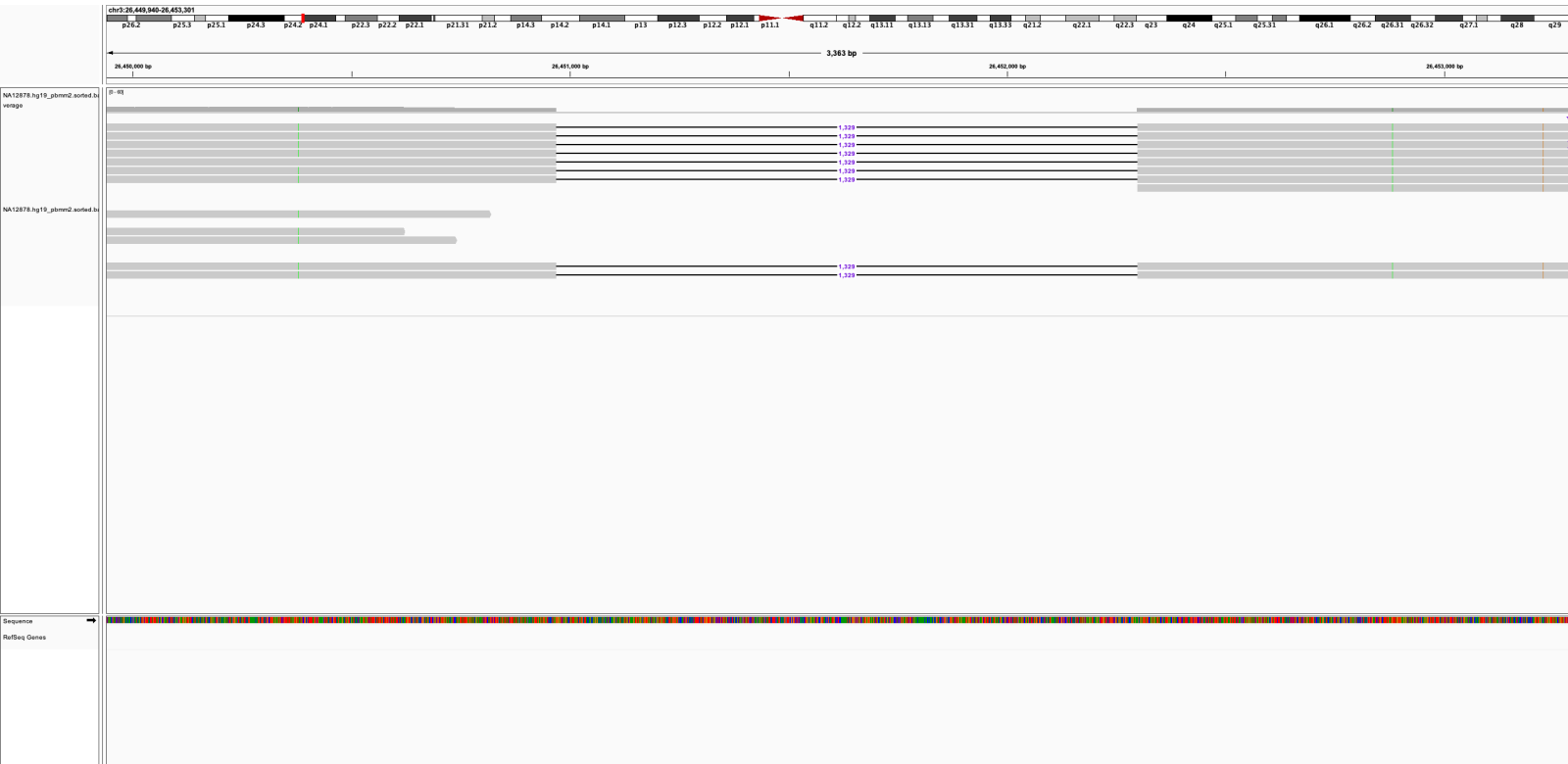


❖ chr2:184,085,515-184,090,985

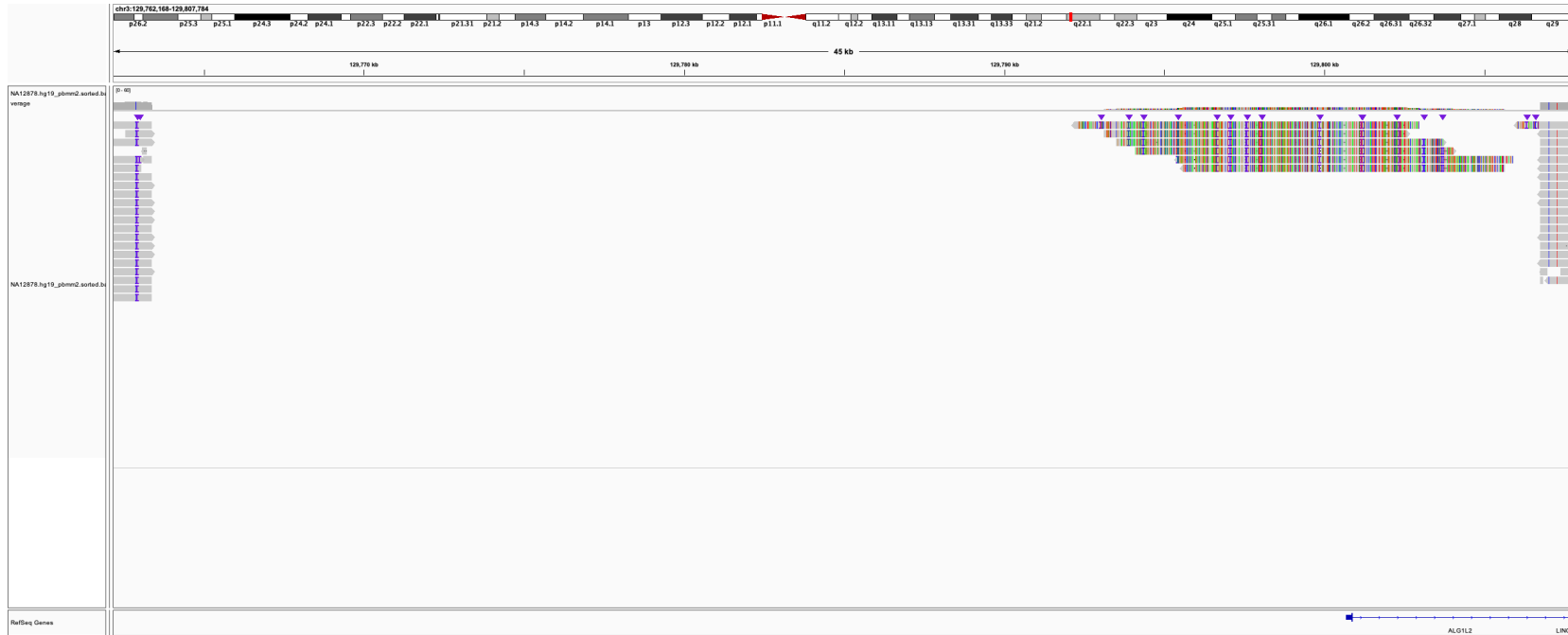




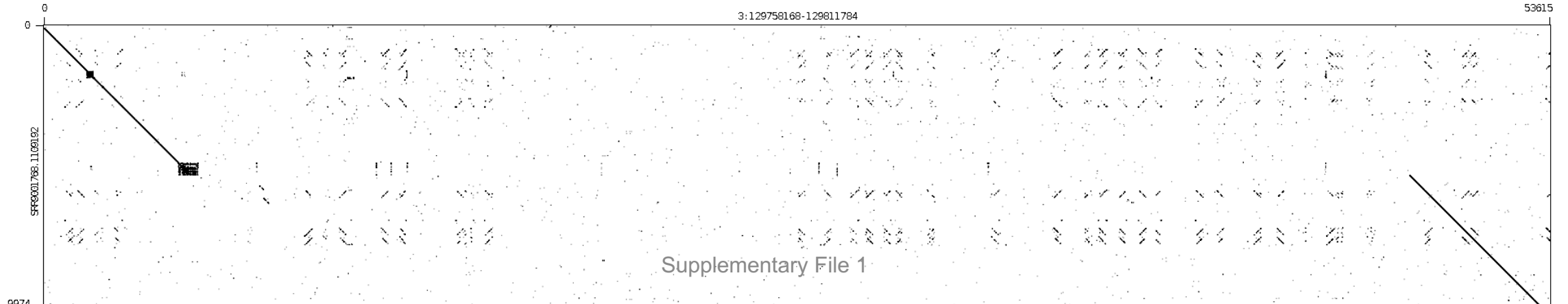
chr3:26,450,940-26,452,301



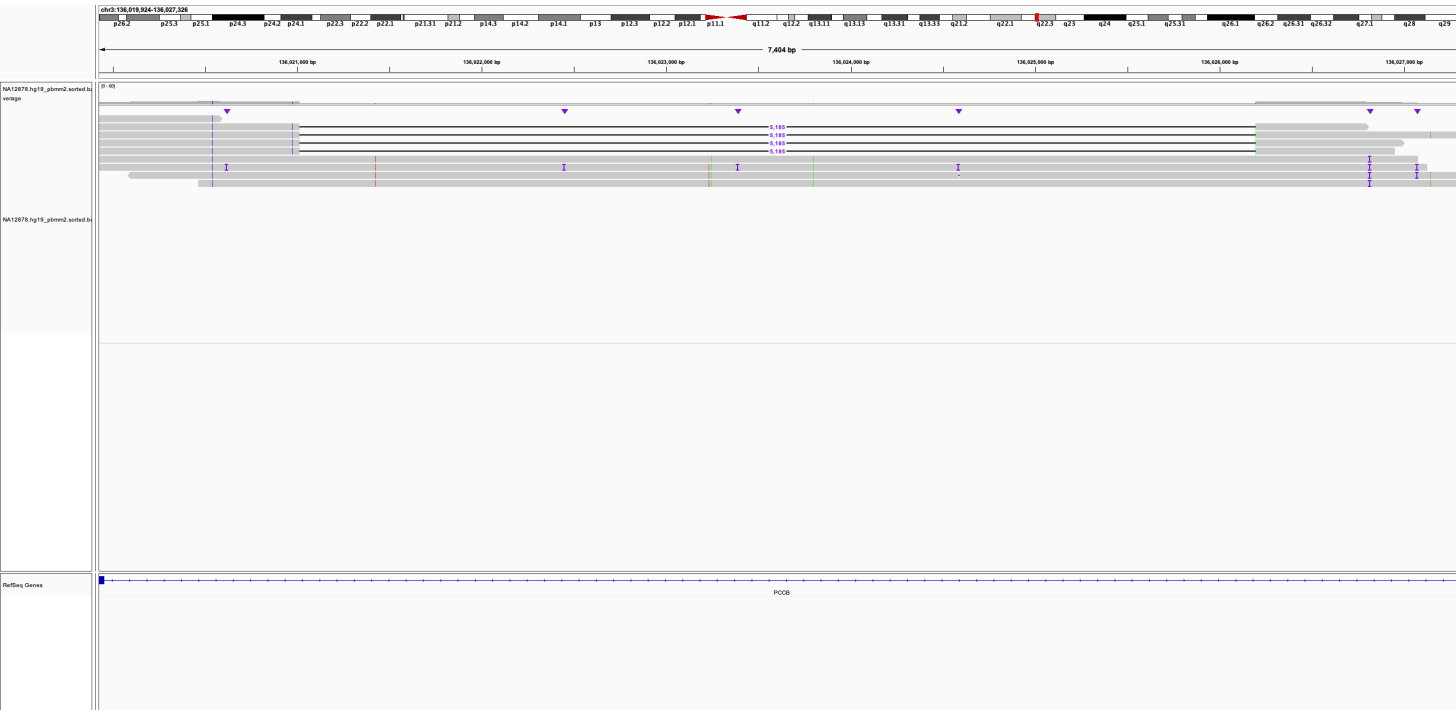
❖ chr3:129,763,168-129,806,784



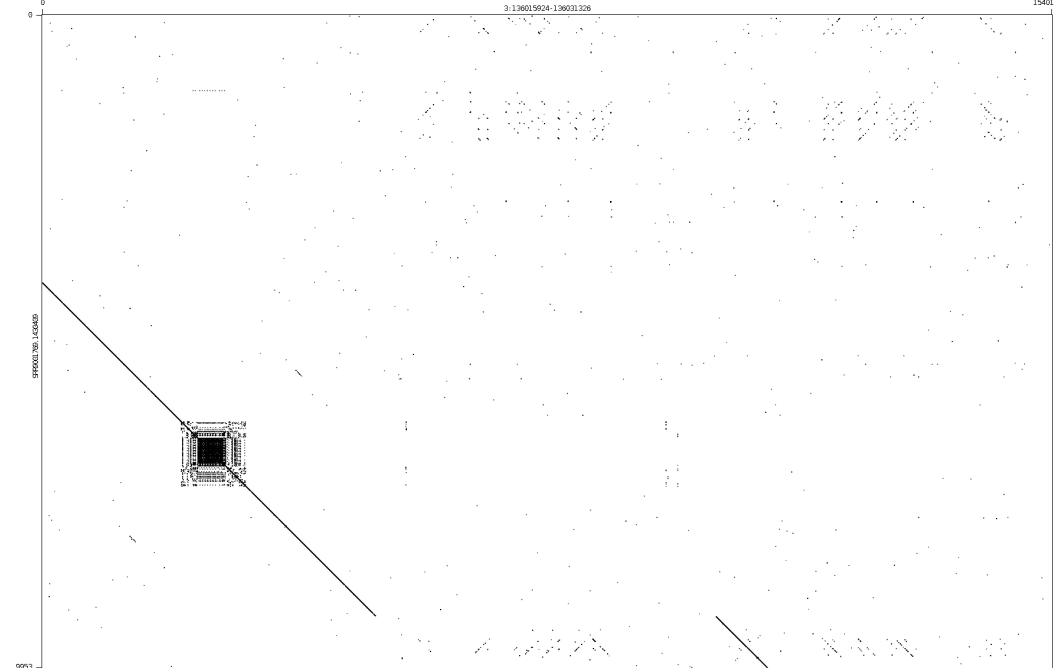
3:129758168-129811784 vs. SRR9001768.1109192
Zoom: 29 : 1
Word length: 10 GC ratio seq1: 0.4687
Window size: 0 GC ratio seq2: 0.4771
Matrix: edna.mat Program: Gepard (1.40 final)



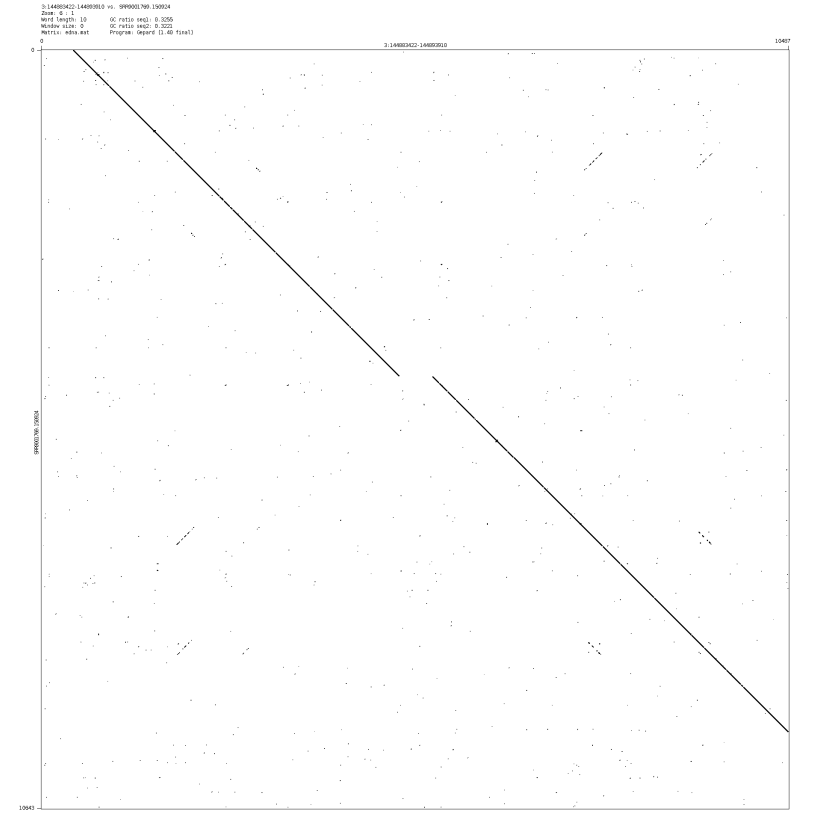
❖ chr3:136,020,924-136,026,326



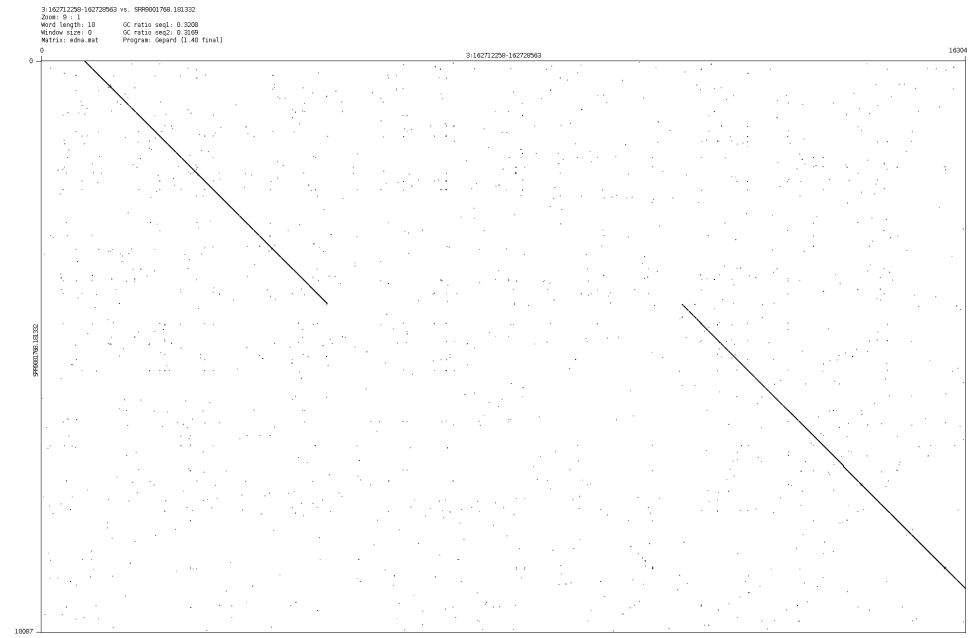
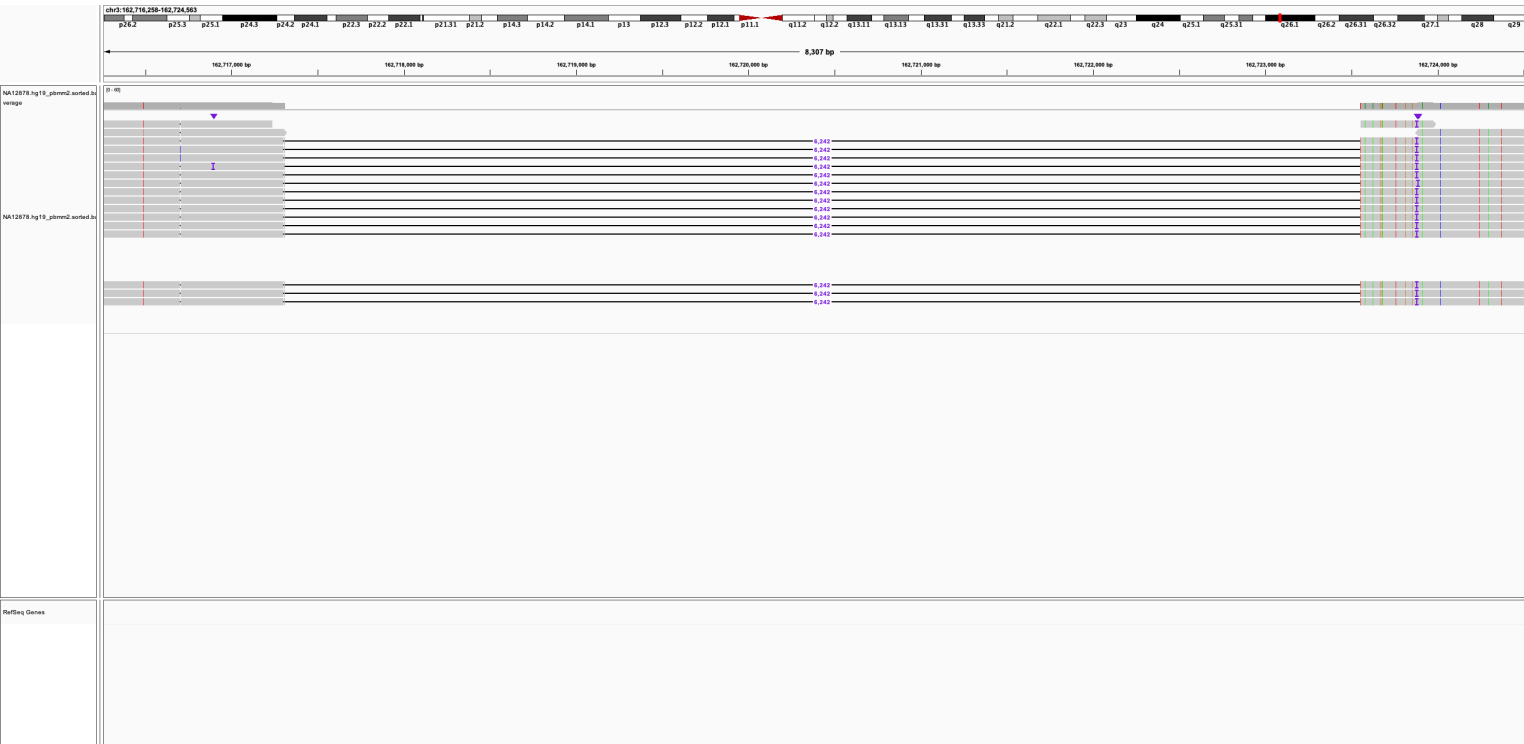
3:136015624-136031326 vs. 599001709-1450409
Zoom: 9 x 1
Word length: 10 GC ratio seq1: 0.4187
Window size: 0 GC ratio seq2: 0.4176
Matrix: edna.mat Program: Gepard (1.40 final)



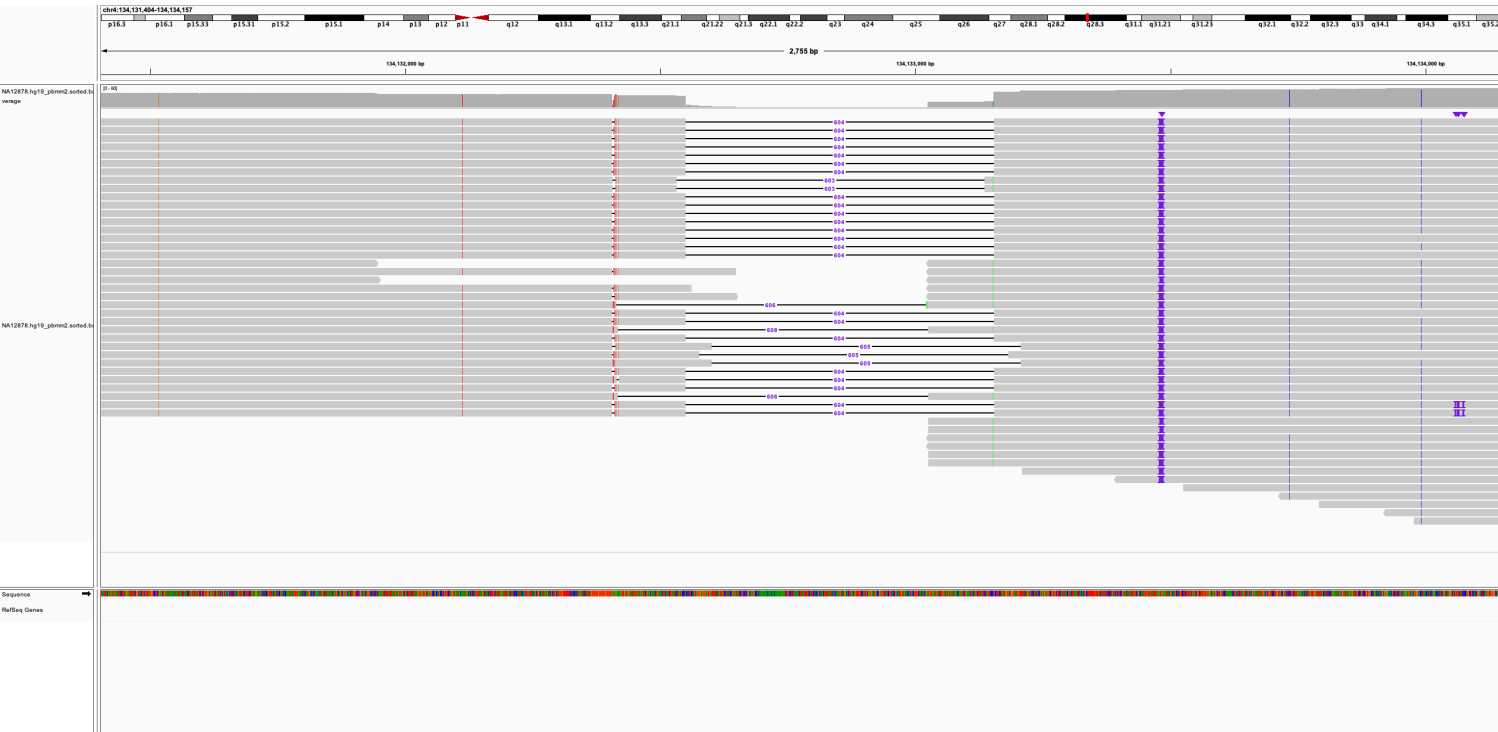
❖ chr3:144,888,422-144,888,910



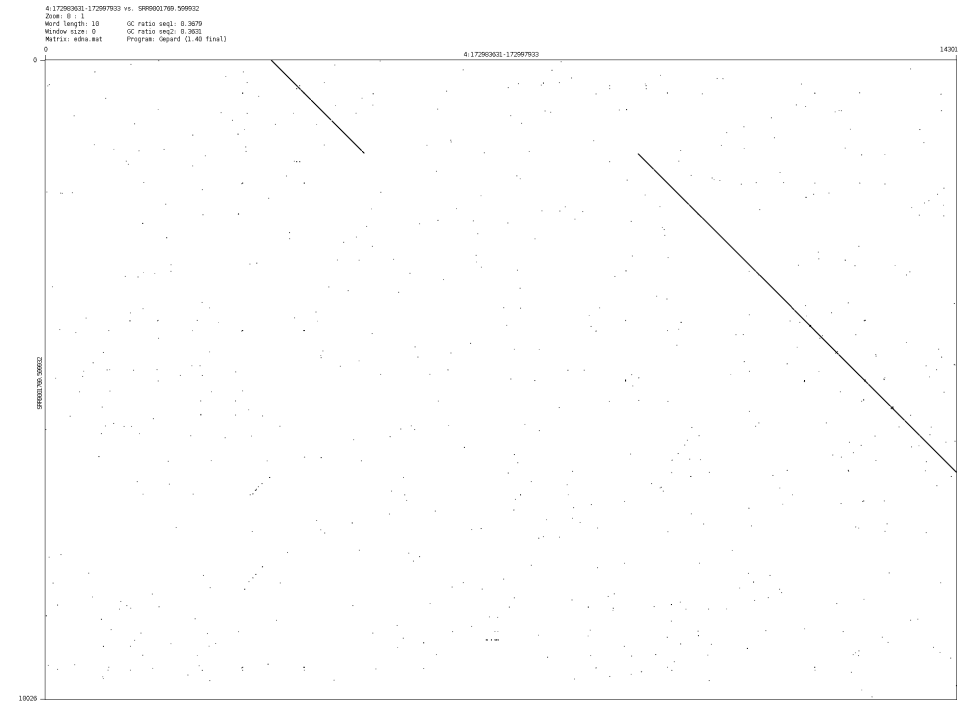
❖ chr3:162,717,258-162,723,563



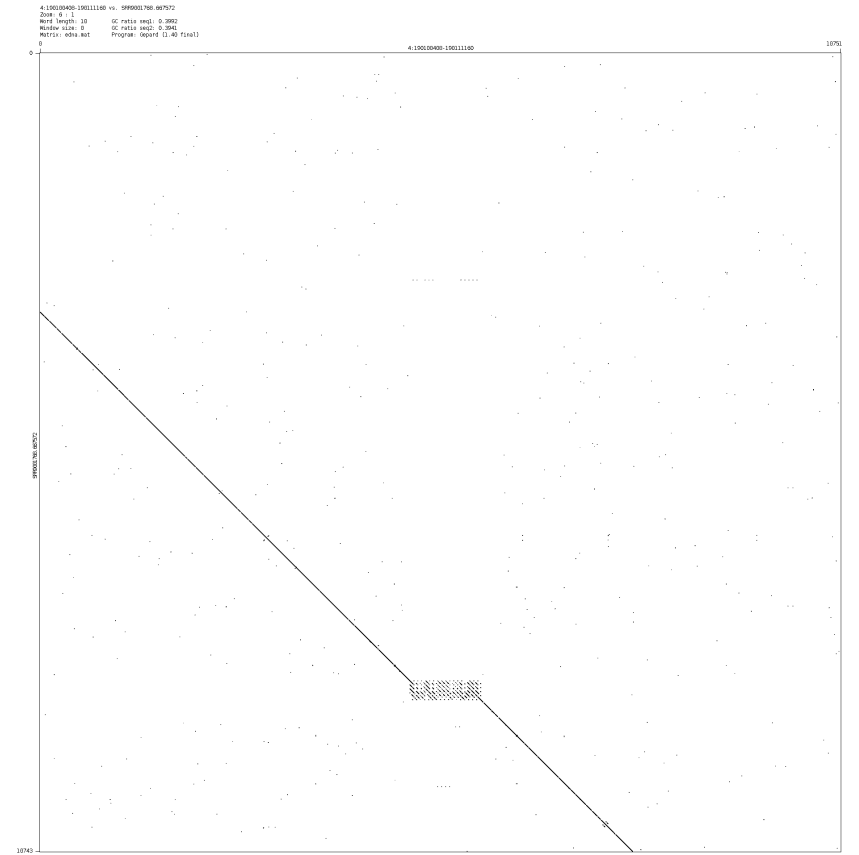
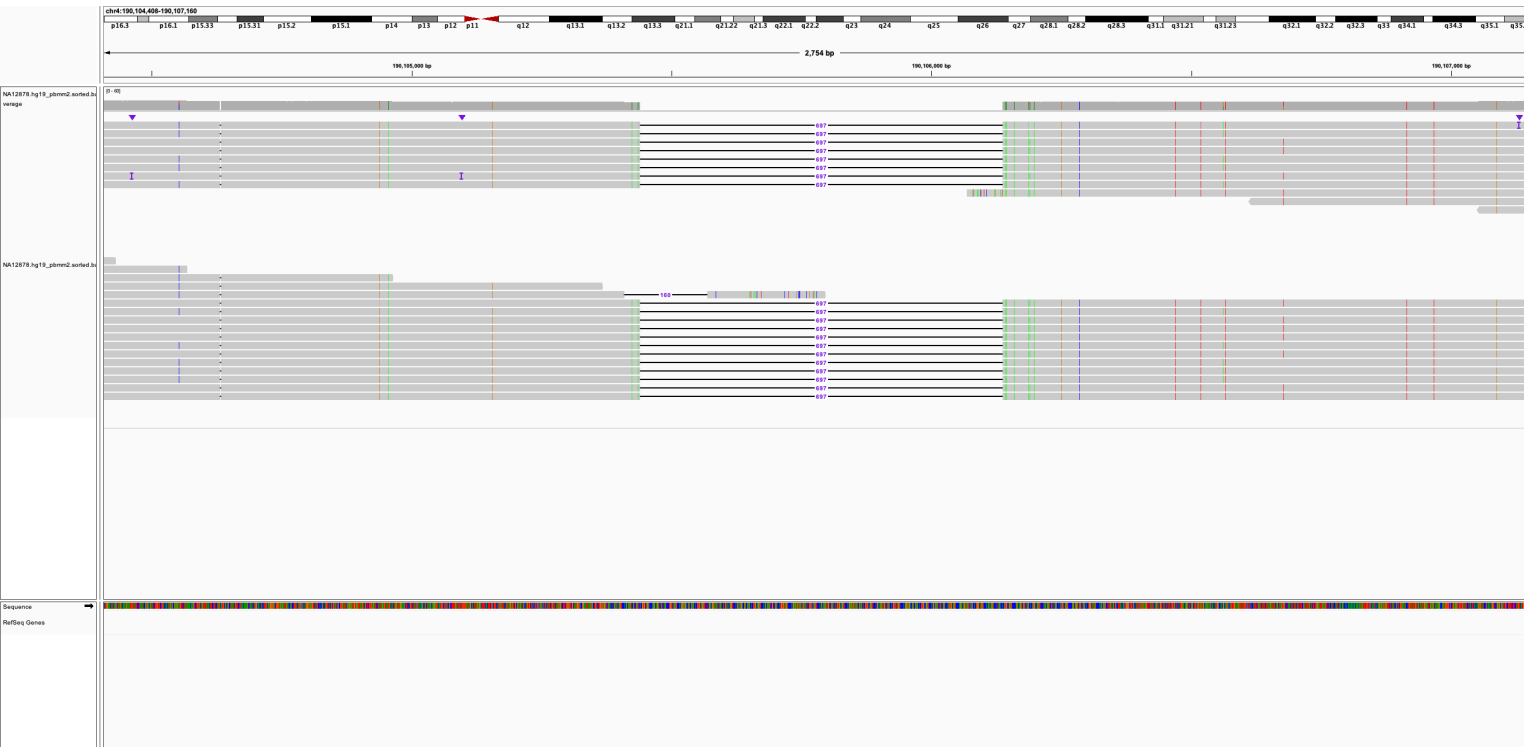
❖ chr4:134,132,404-134,133,157



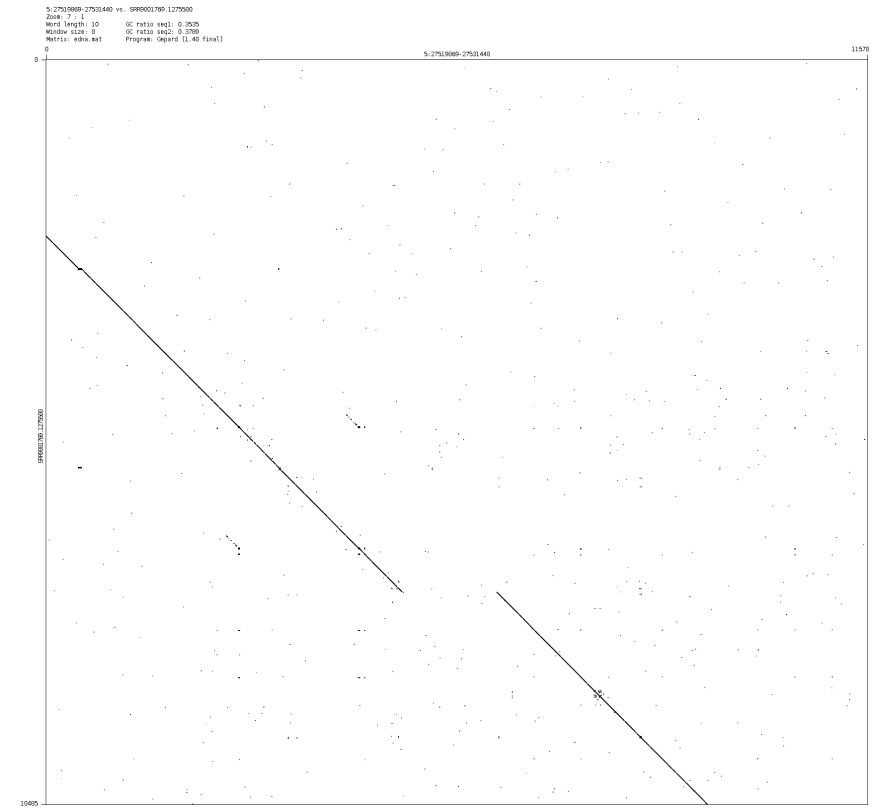
❖ chr4:172,988,631-172,992,933



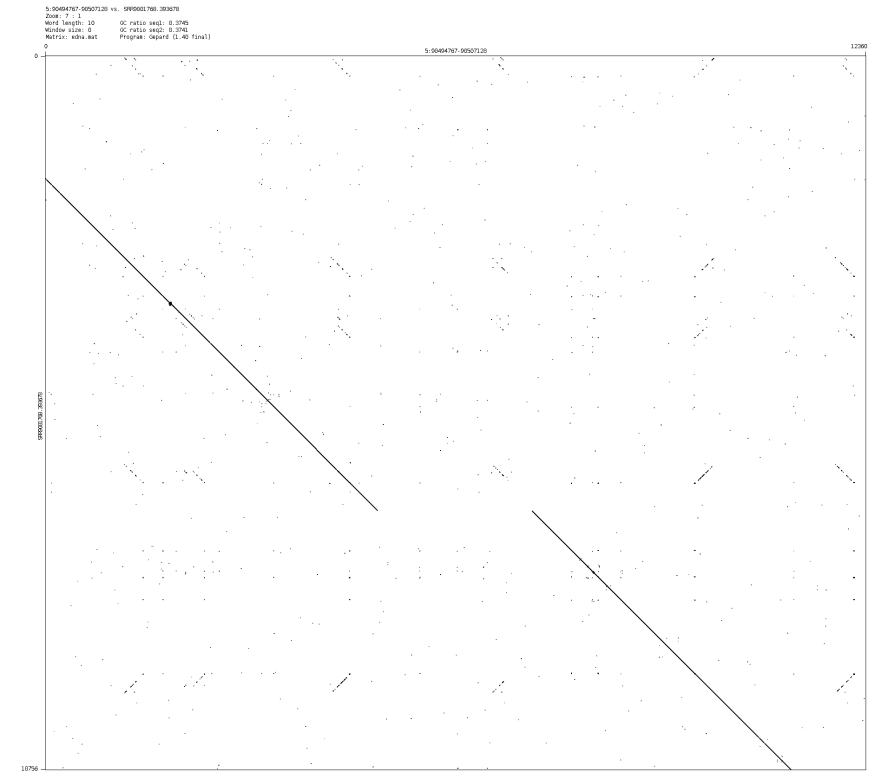
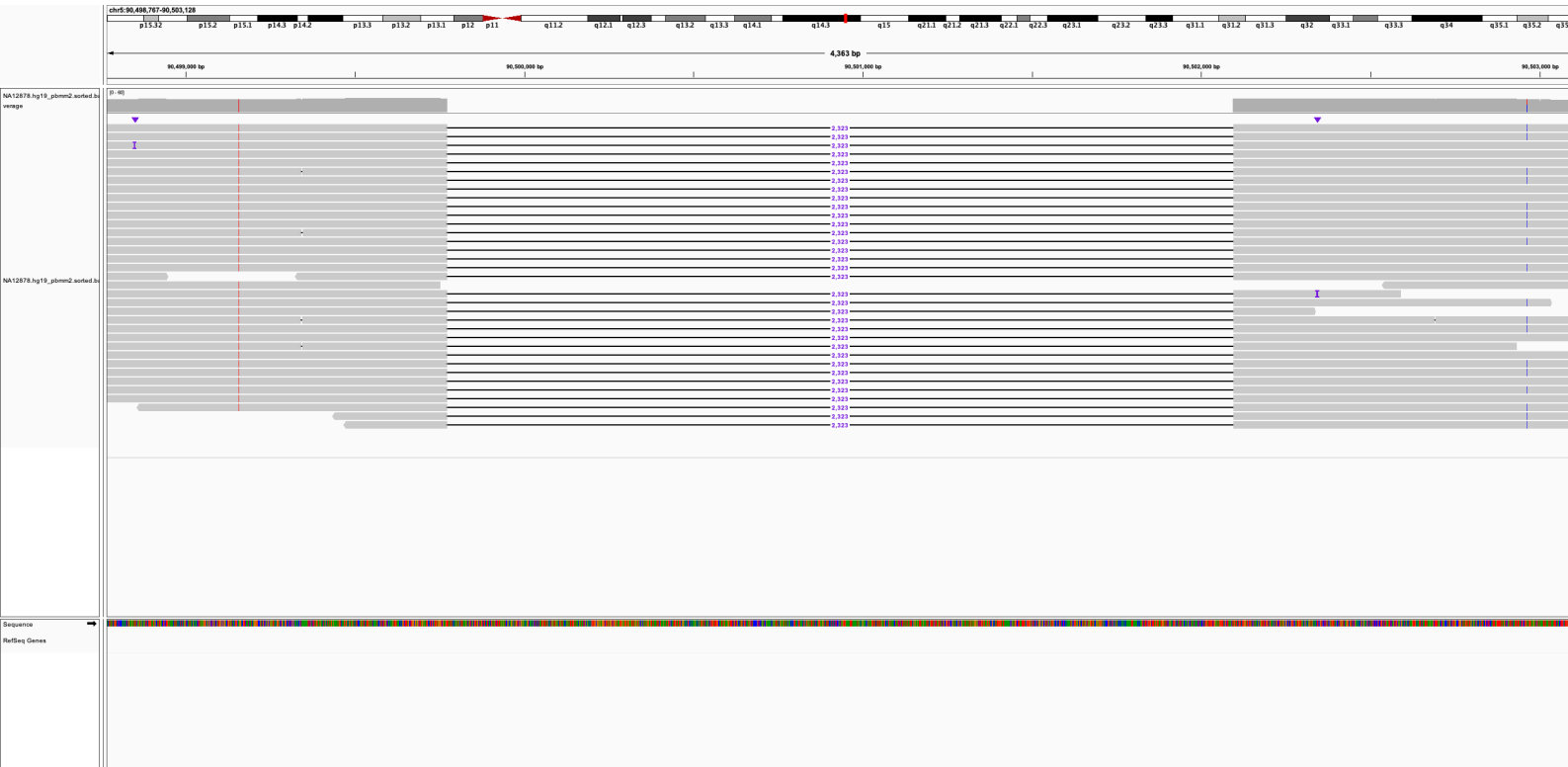
❖ chr4:190,105,408-190,106,160



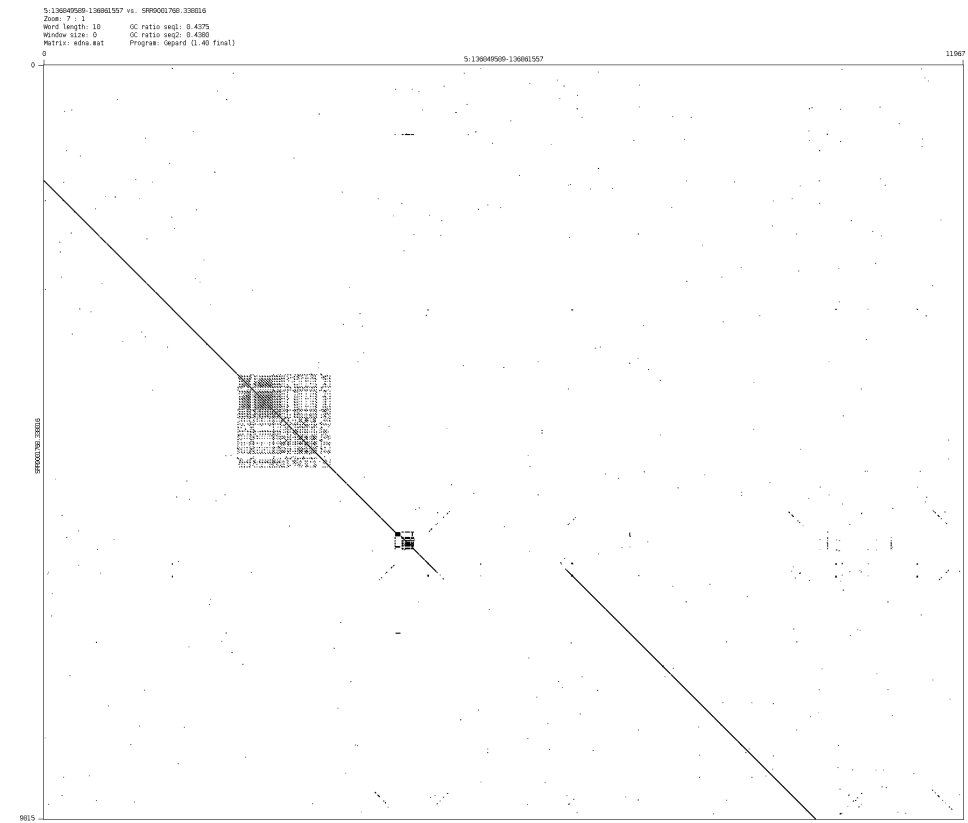
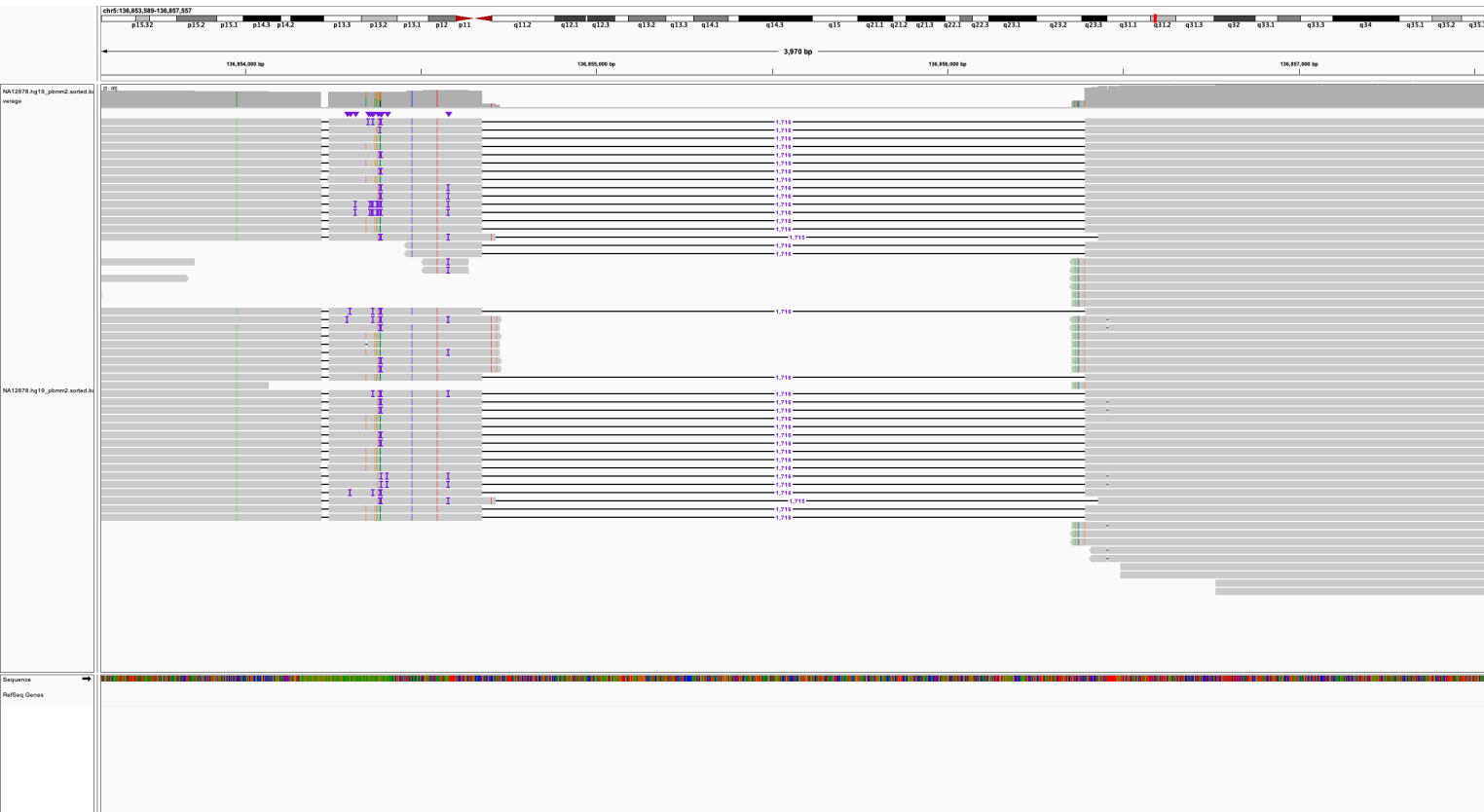
❖ chr5:27,524,869-27,526,440



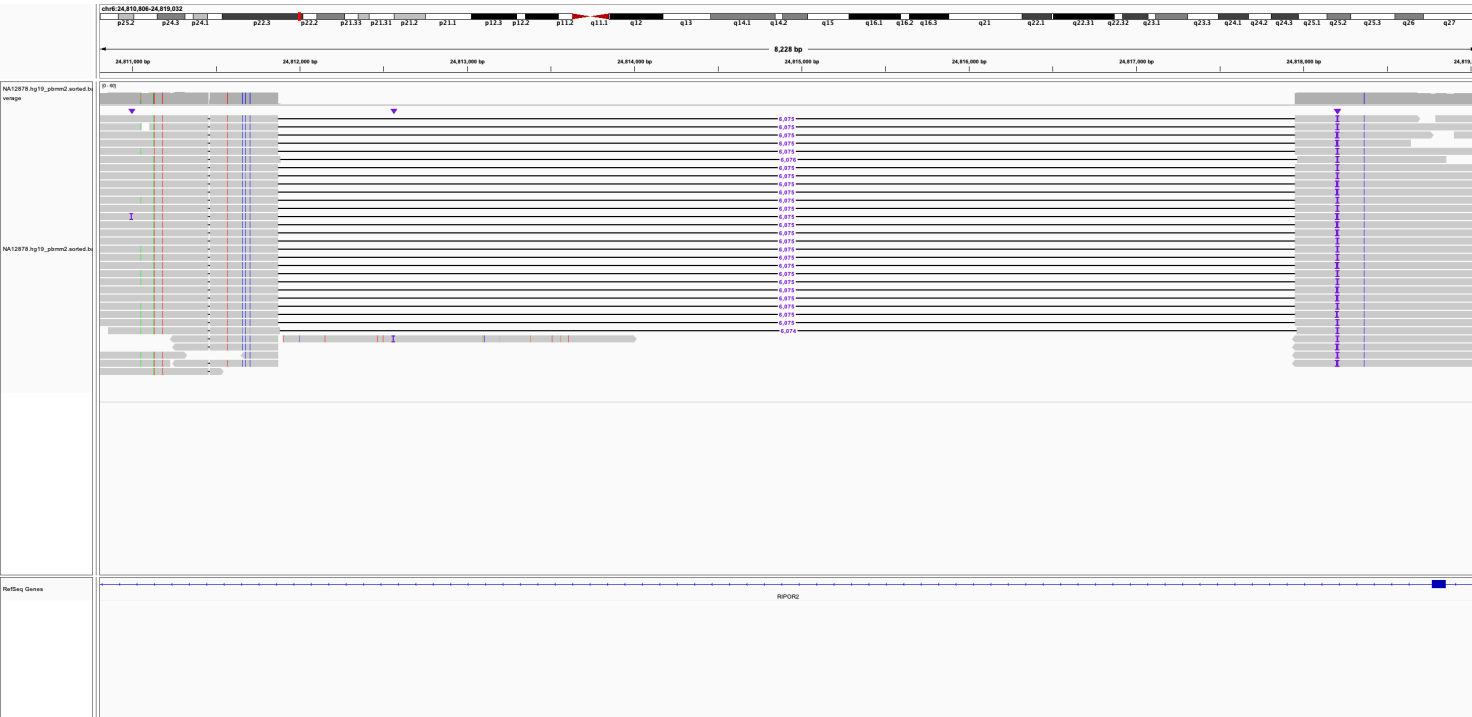
❖ chr5:90,499,767-90,502,128



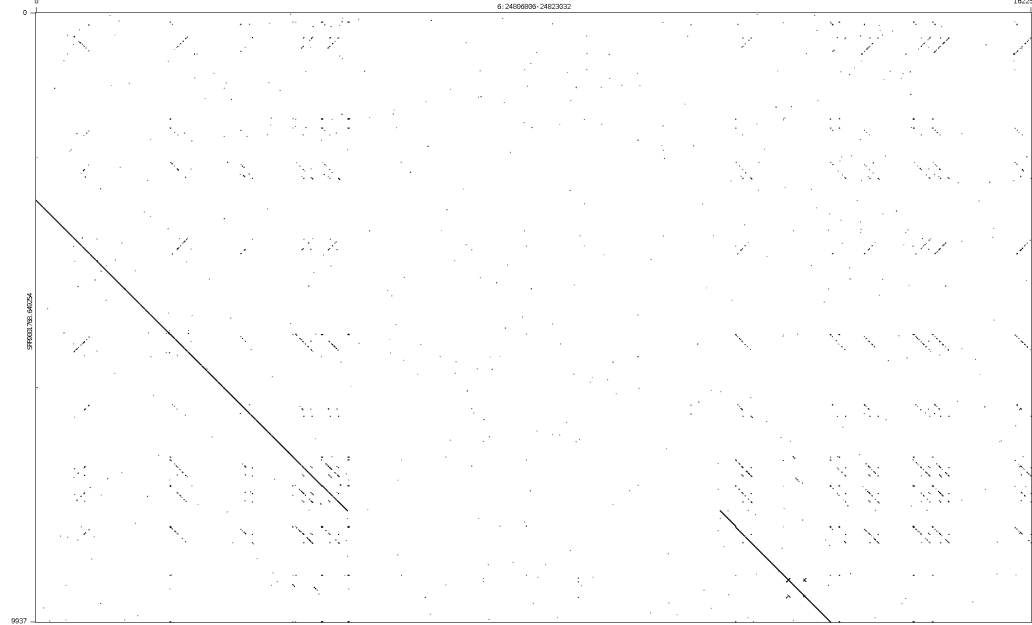
❖ chr5:136,854,589-136,856,557



❖ chr6:24,811,806-24,818,032

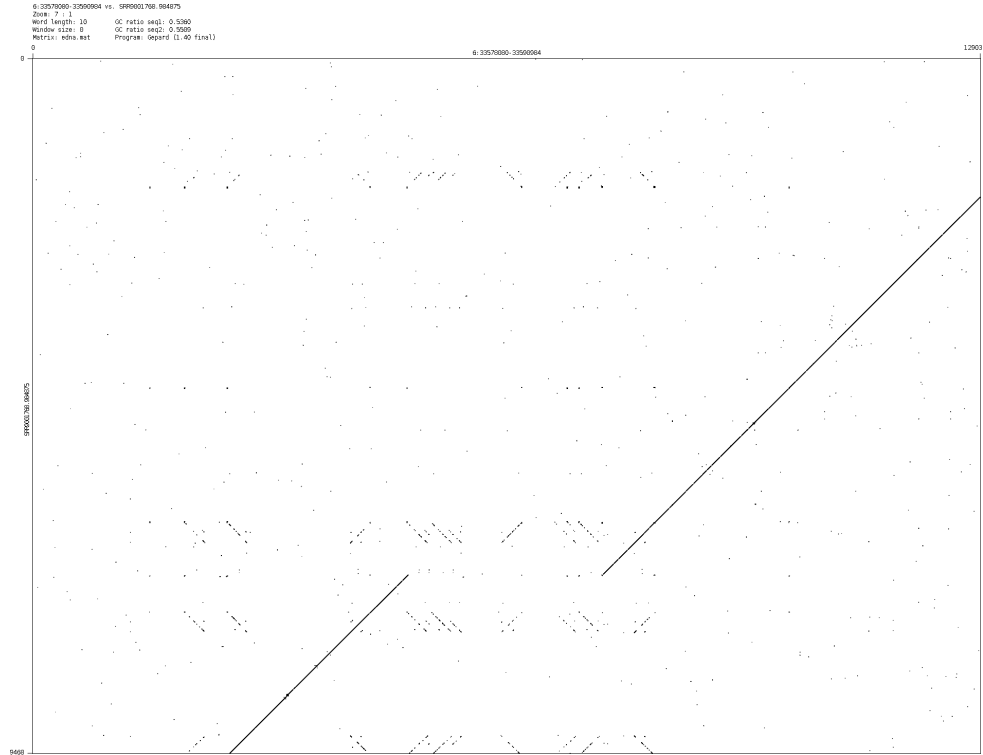


6:24808906-24823032 vs. 599001766-600254
Zoom: 9 x 1 GC ratio seq1: 0.4200
Window length: 10 GC ratio seq2: 0.4048
Window size: 0
Matrix: edna-wat Program: Gepard (1.40 final)

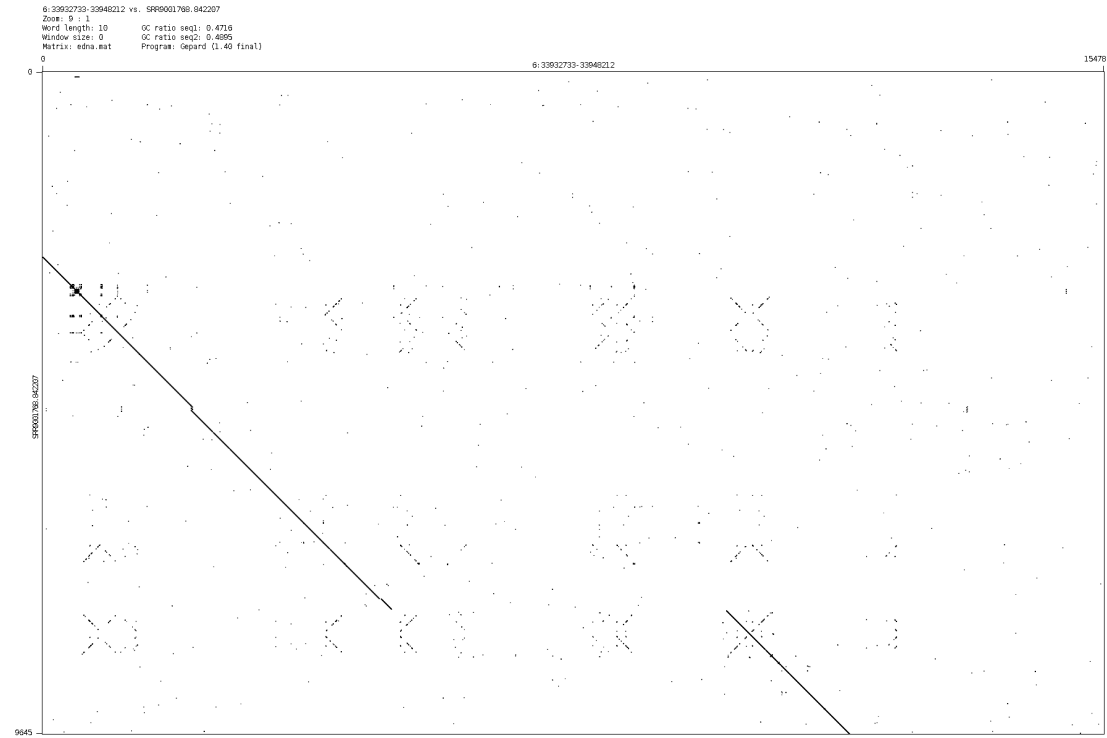




chr6:33,583,080-33,585,984

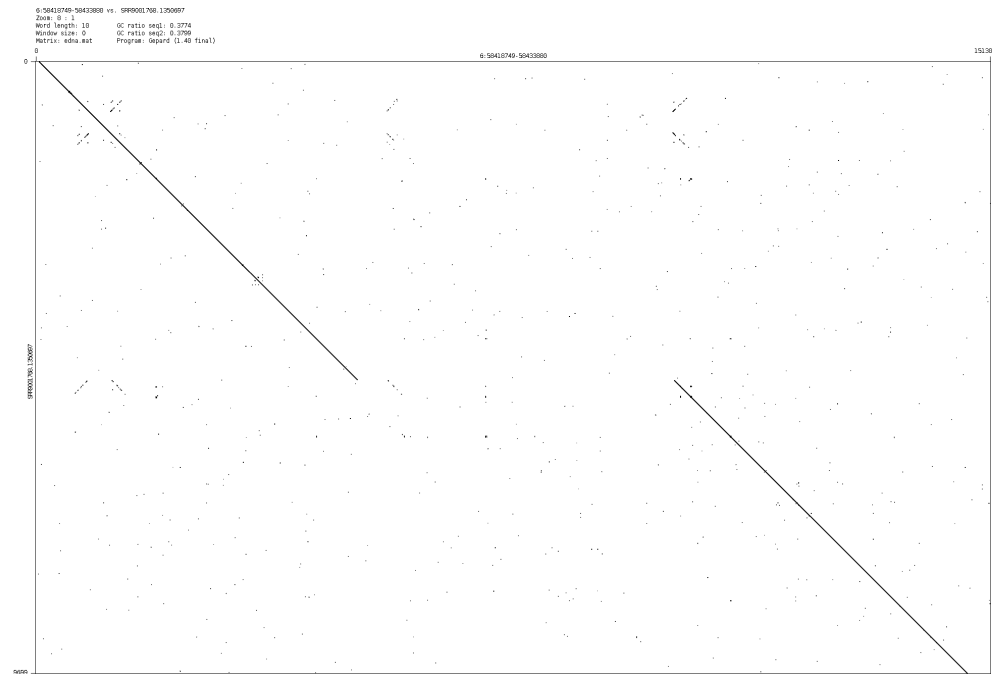


❖ chr6:33,937,733-33,943,212

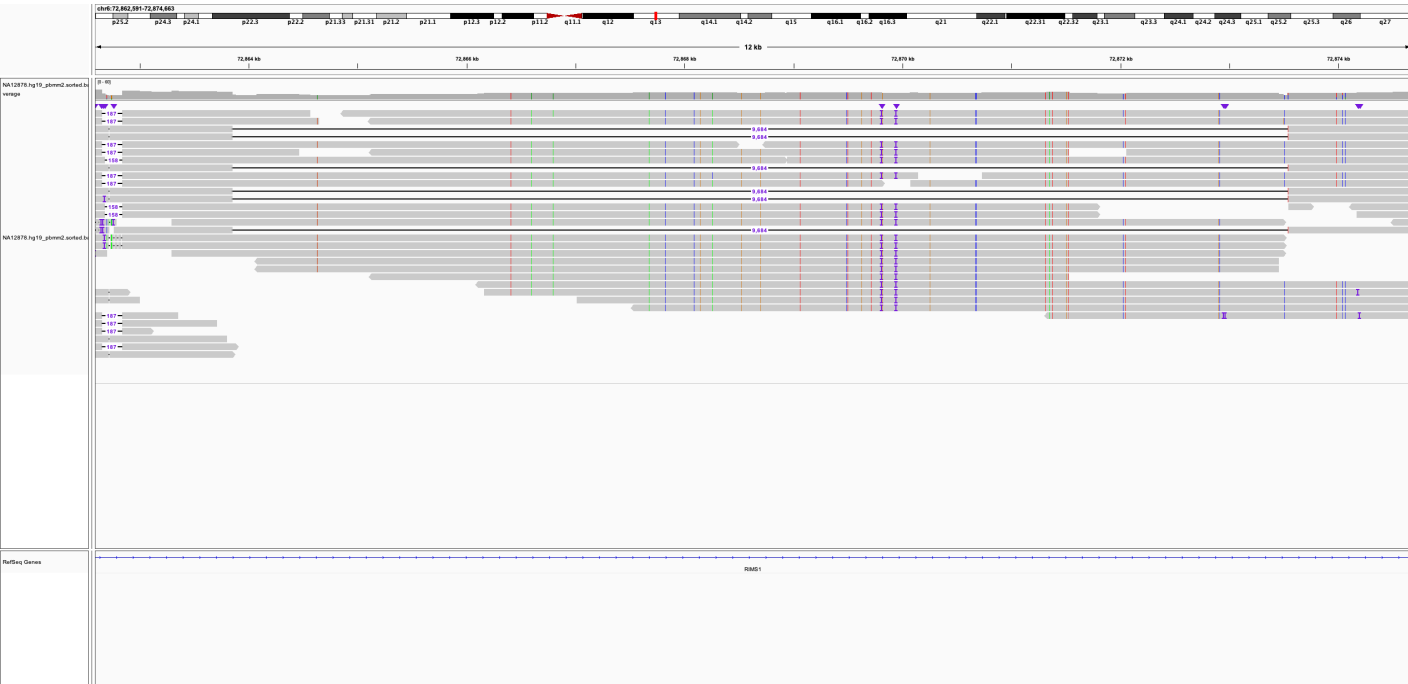




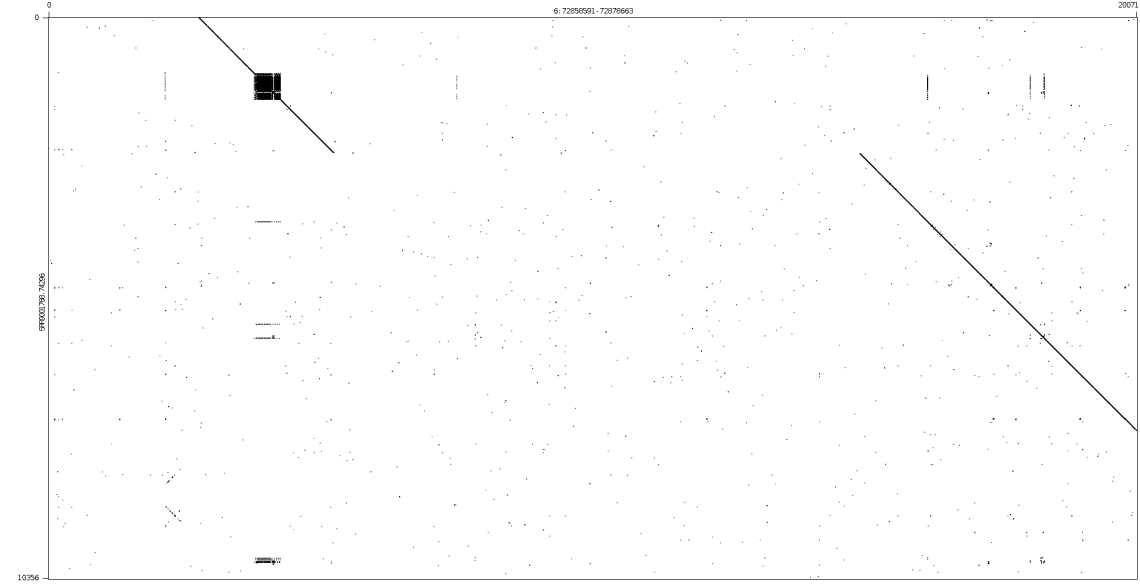
chr6:58,423,749-58,428,880



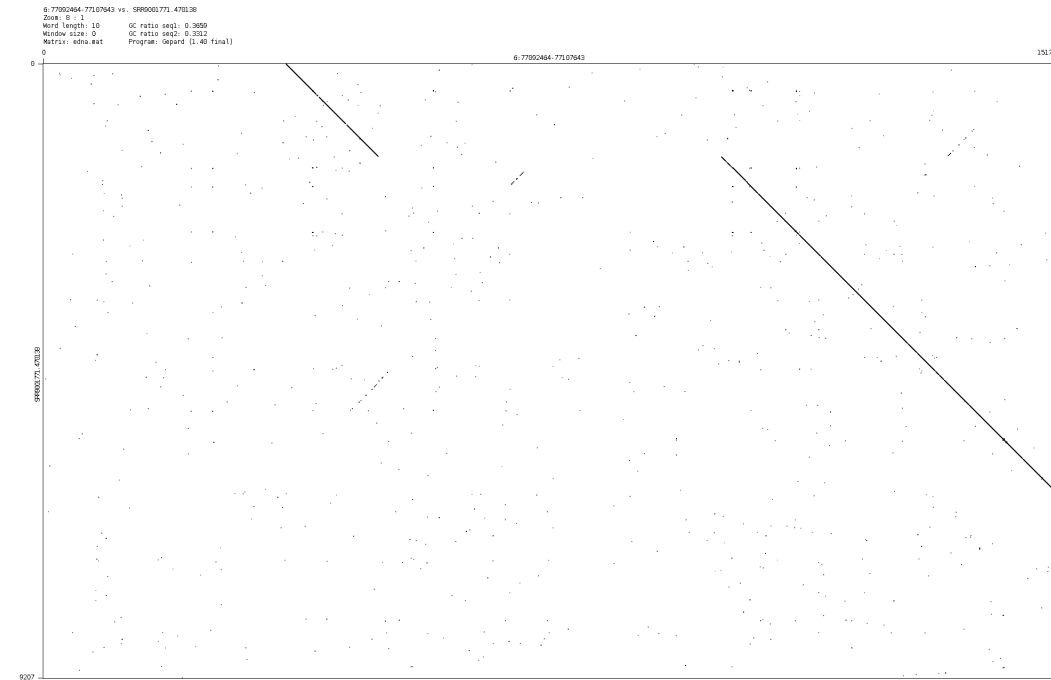
❖ chr6:72,863,591-72,873,663



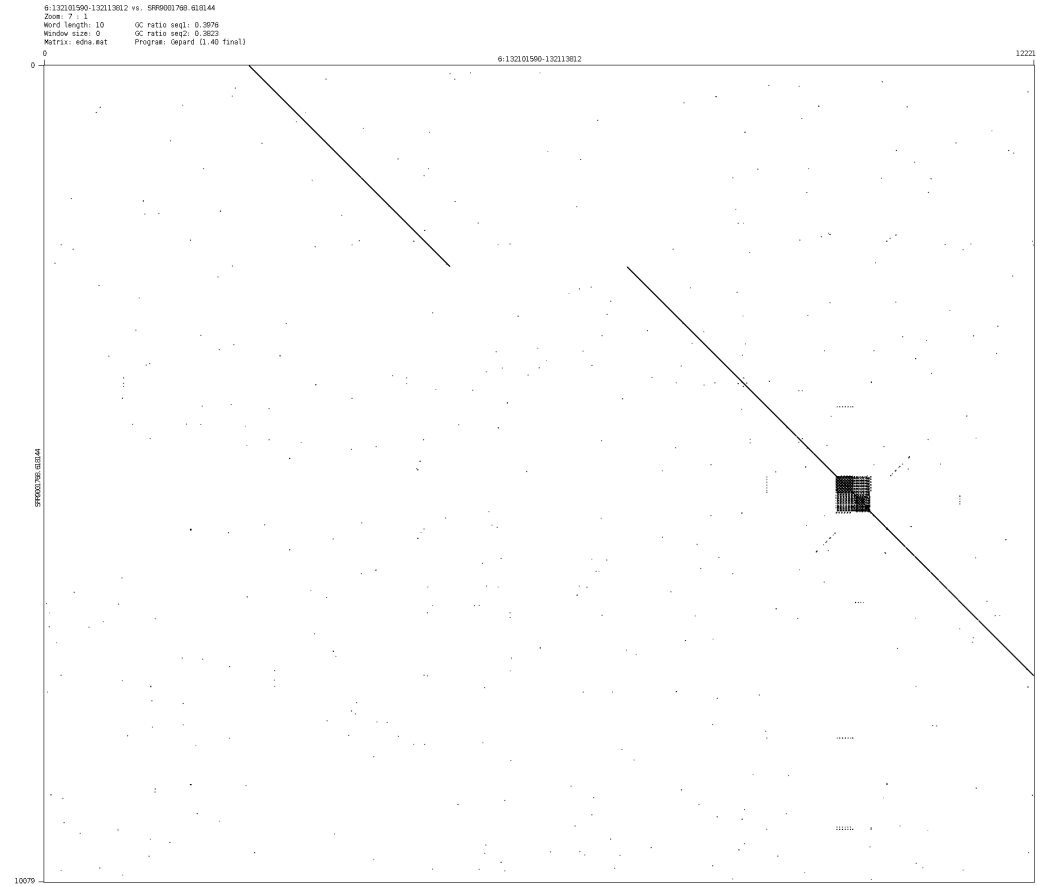
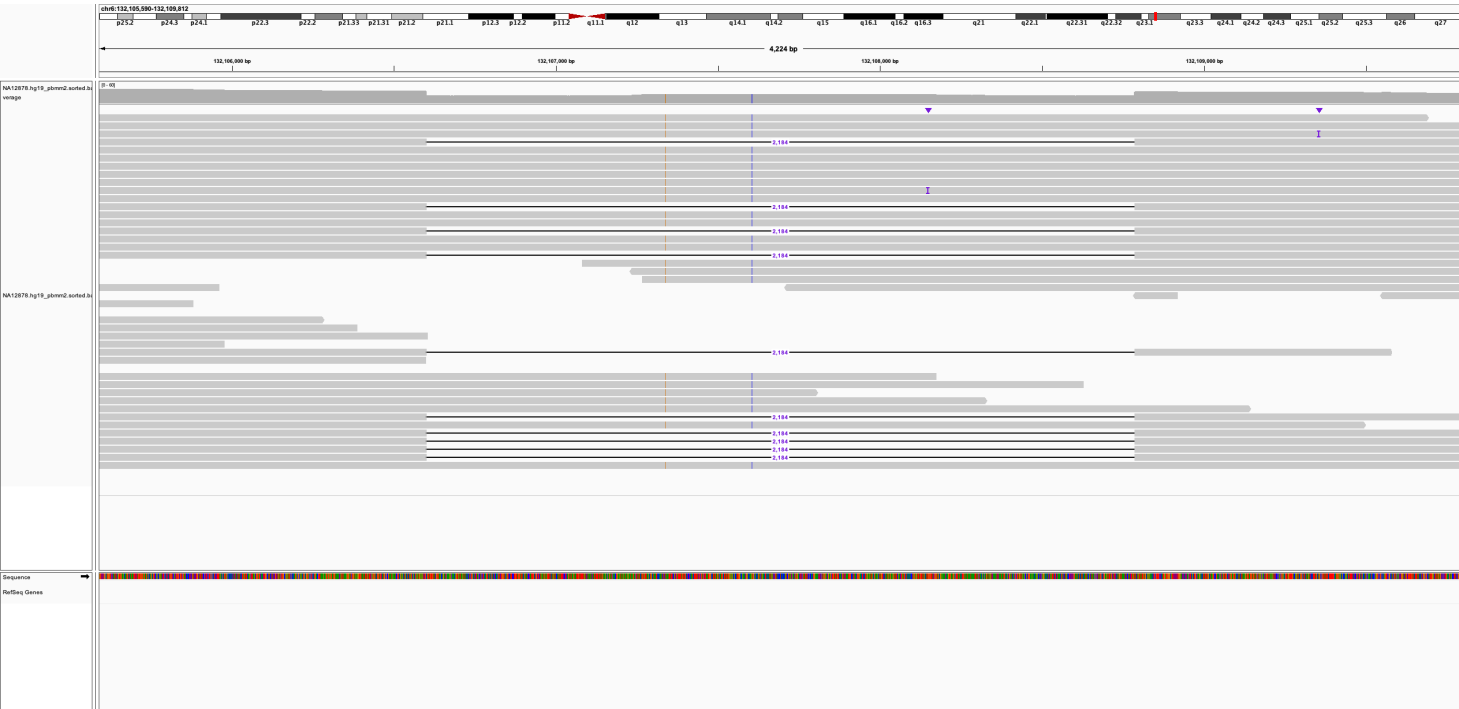
6:72859591-72876663 vs. SRR6001760.74296
Zoom: 11 | 1 GC ratio seq1: 0.3761
Word length: 10 GC ratio seq2: 0.3551
Window size: 0 GC ratio seq3: 0.3551
Matrix: edna.mat Program: Gepard (1.40 final)



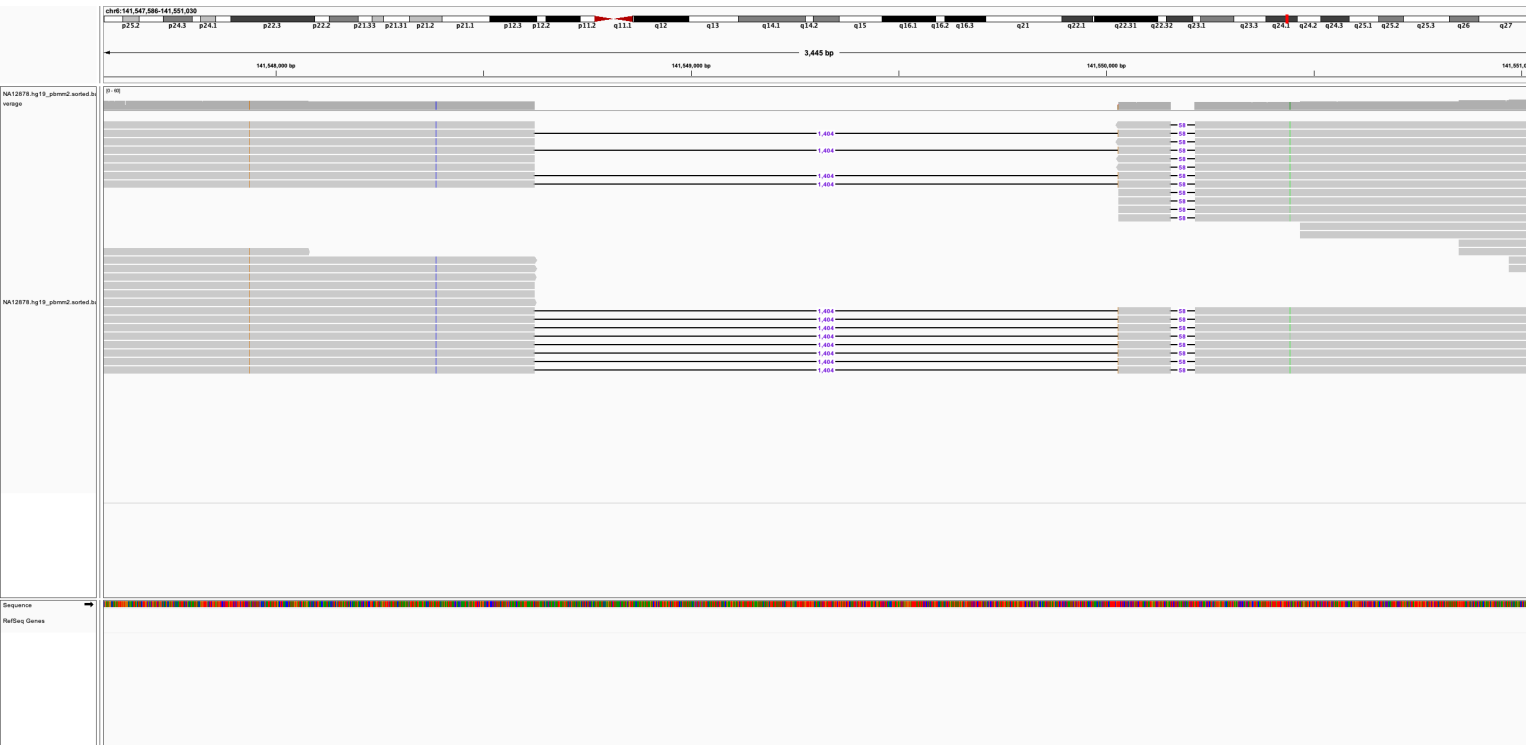
❖ chr6:77,097,464-77,102,643



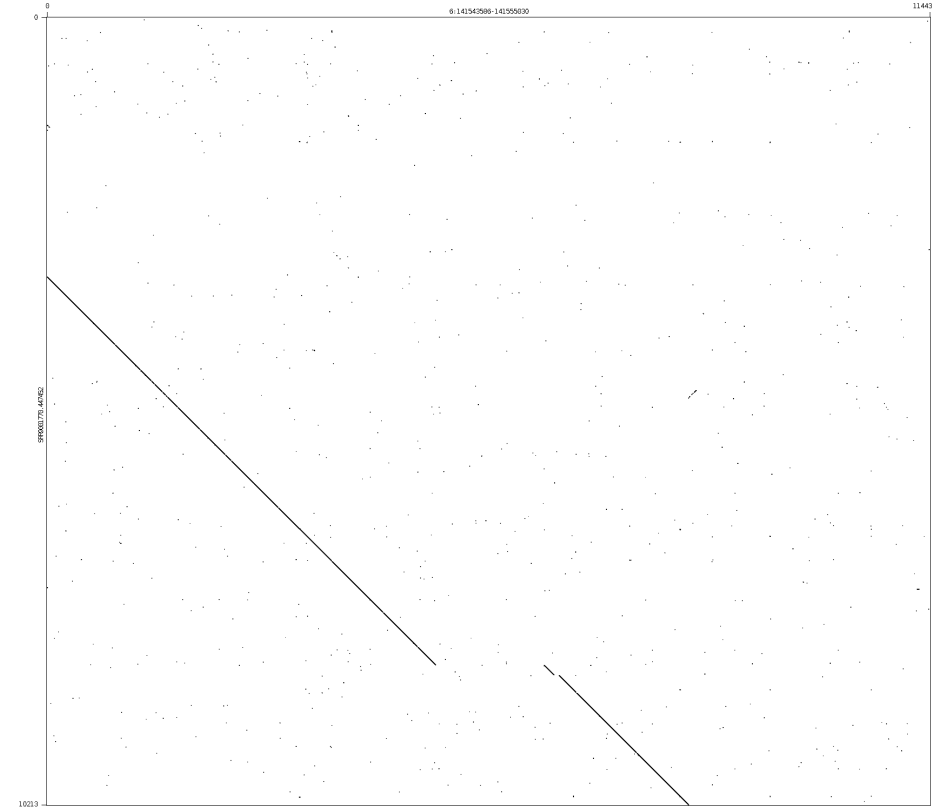
❖ chr6:132,106,590-132,108,812



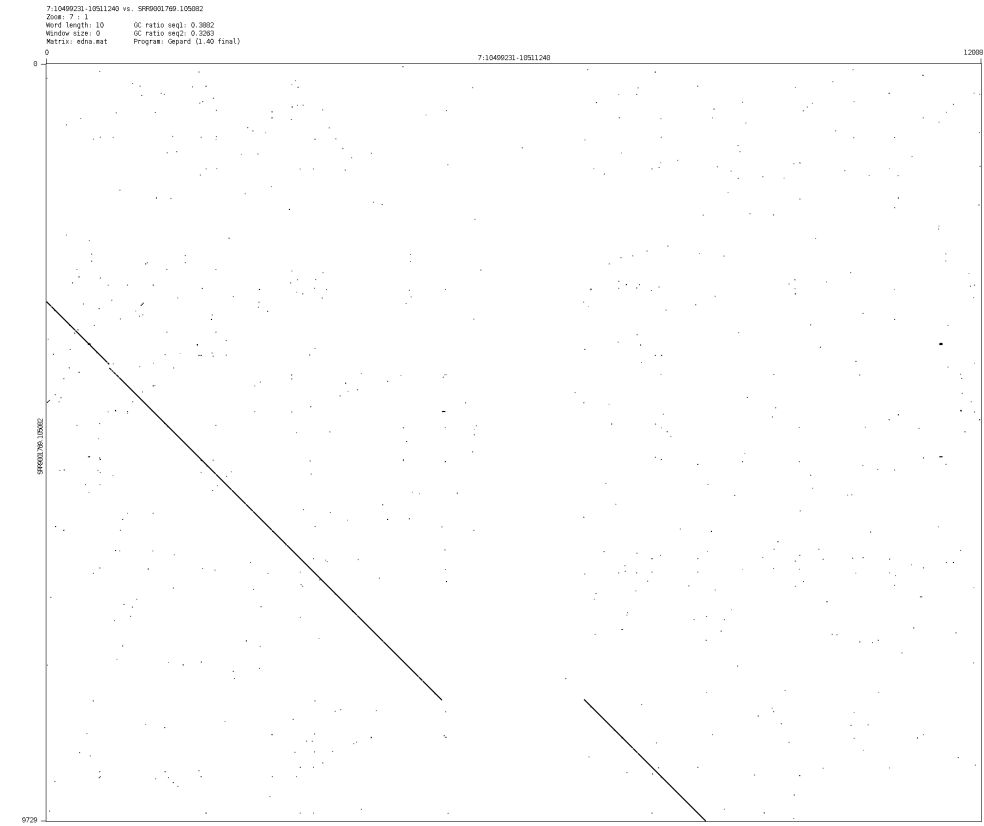
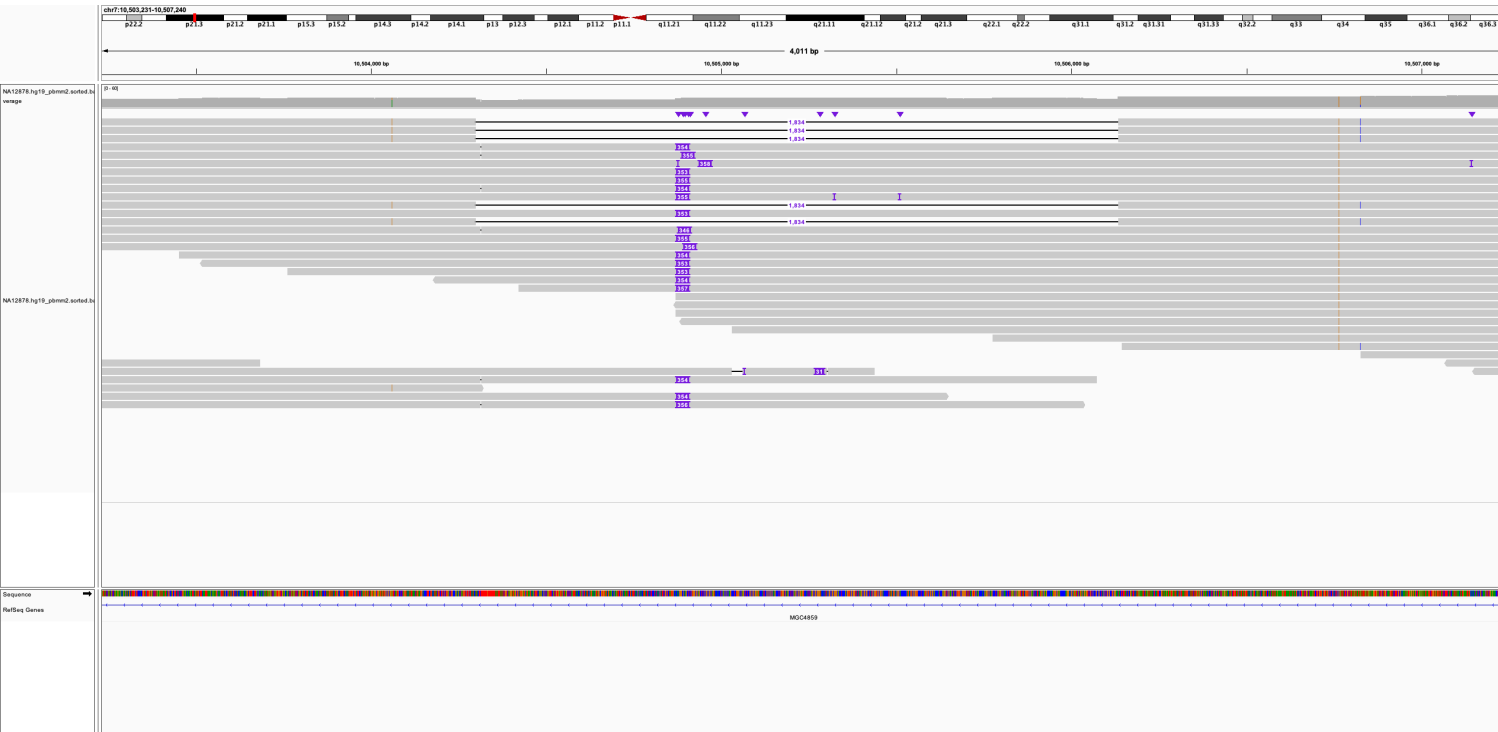
❖ chr6:141,548,586-141,550,030



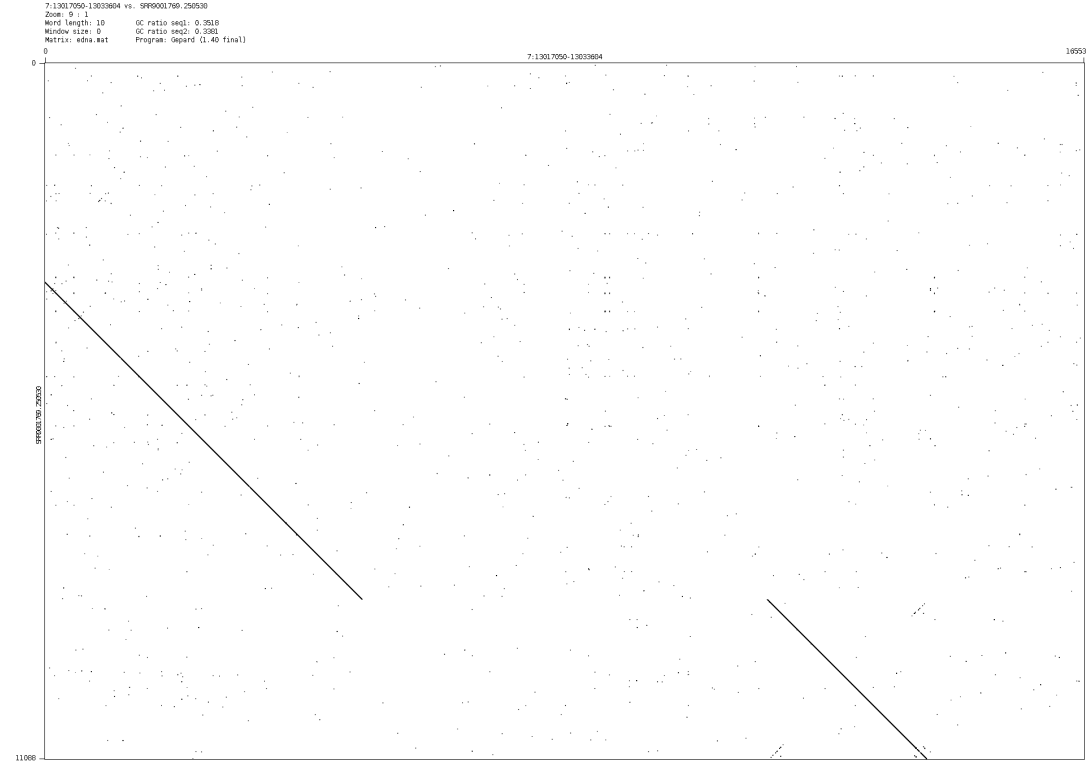
6:141549590-141550030 vs. 999001770.447952
Zoom: 7 x 1
word length: 30 GC ratio 9921: 0.3348
window size: 0 GC ratio 9922: 0.3413
Matrix: kma.mat Program: Gapped (L) 93 final1



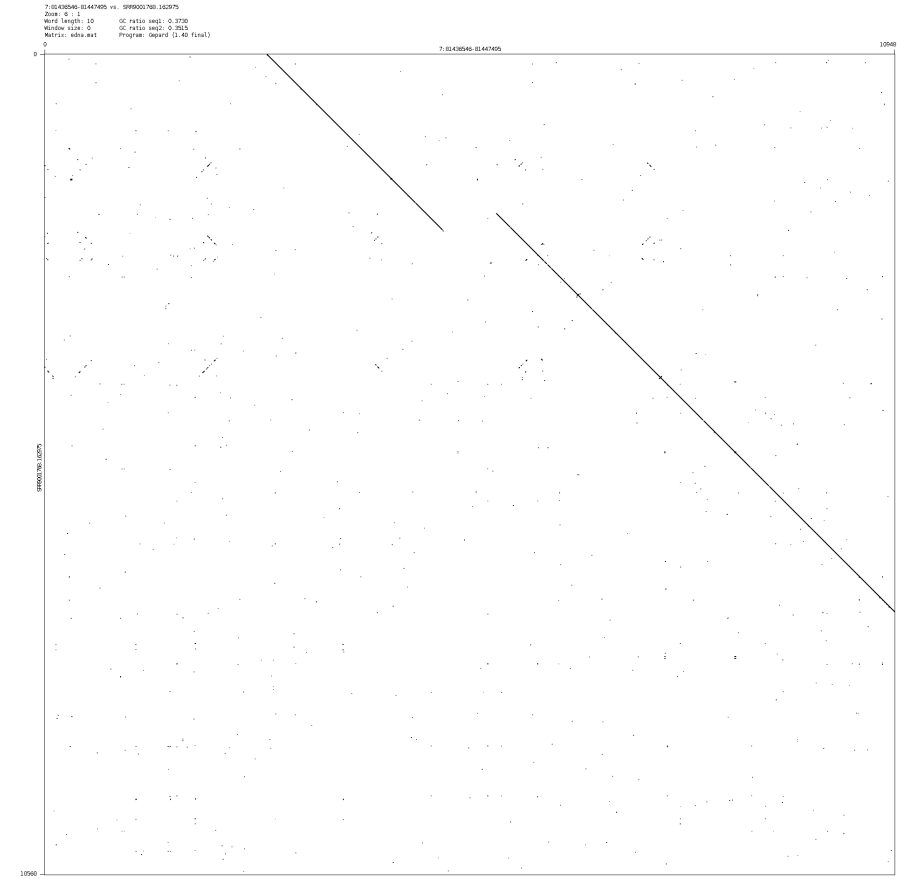
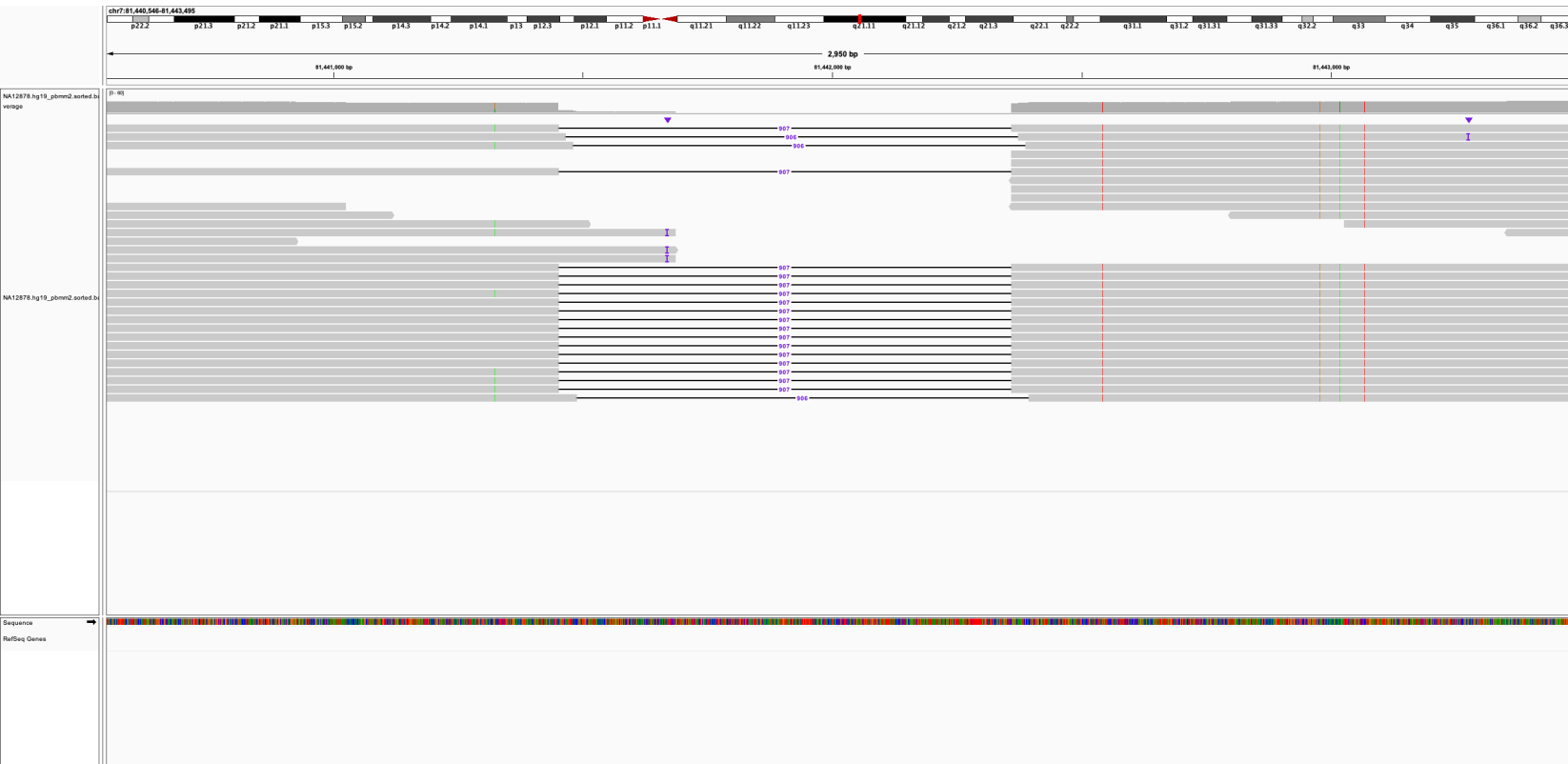
❖ chr7:10,504,231-10,506,240



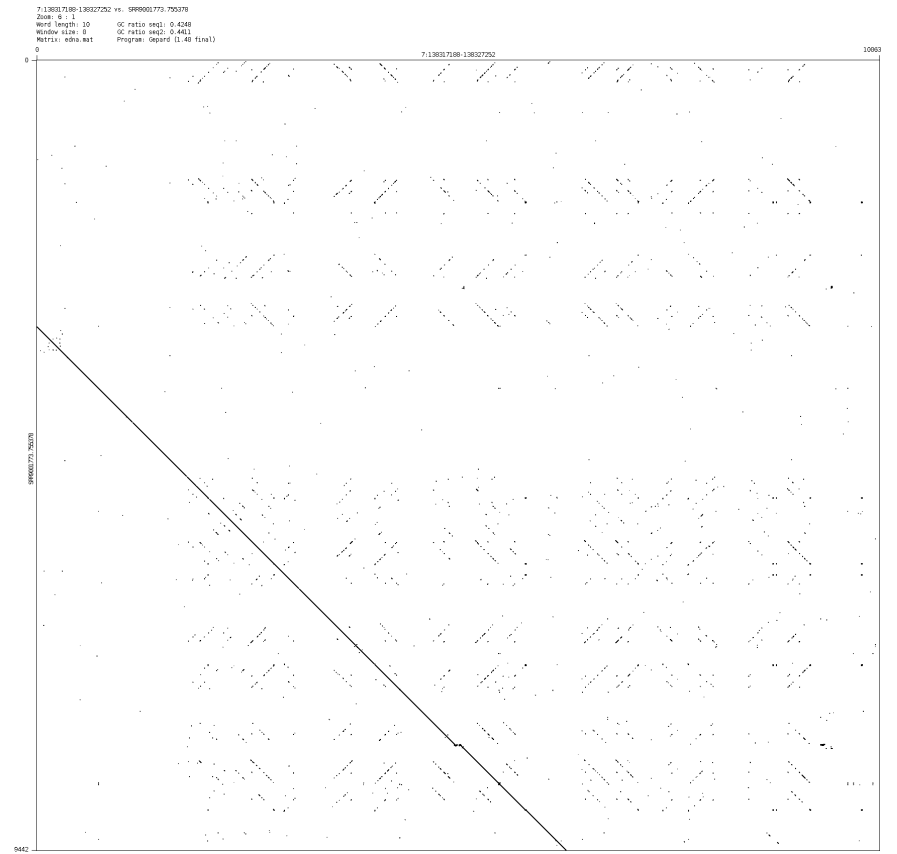
❖ chr7:13,022,050-13,028,604



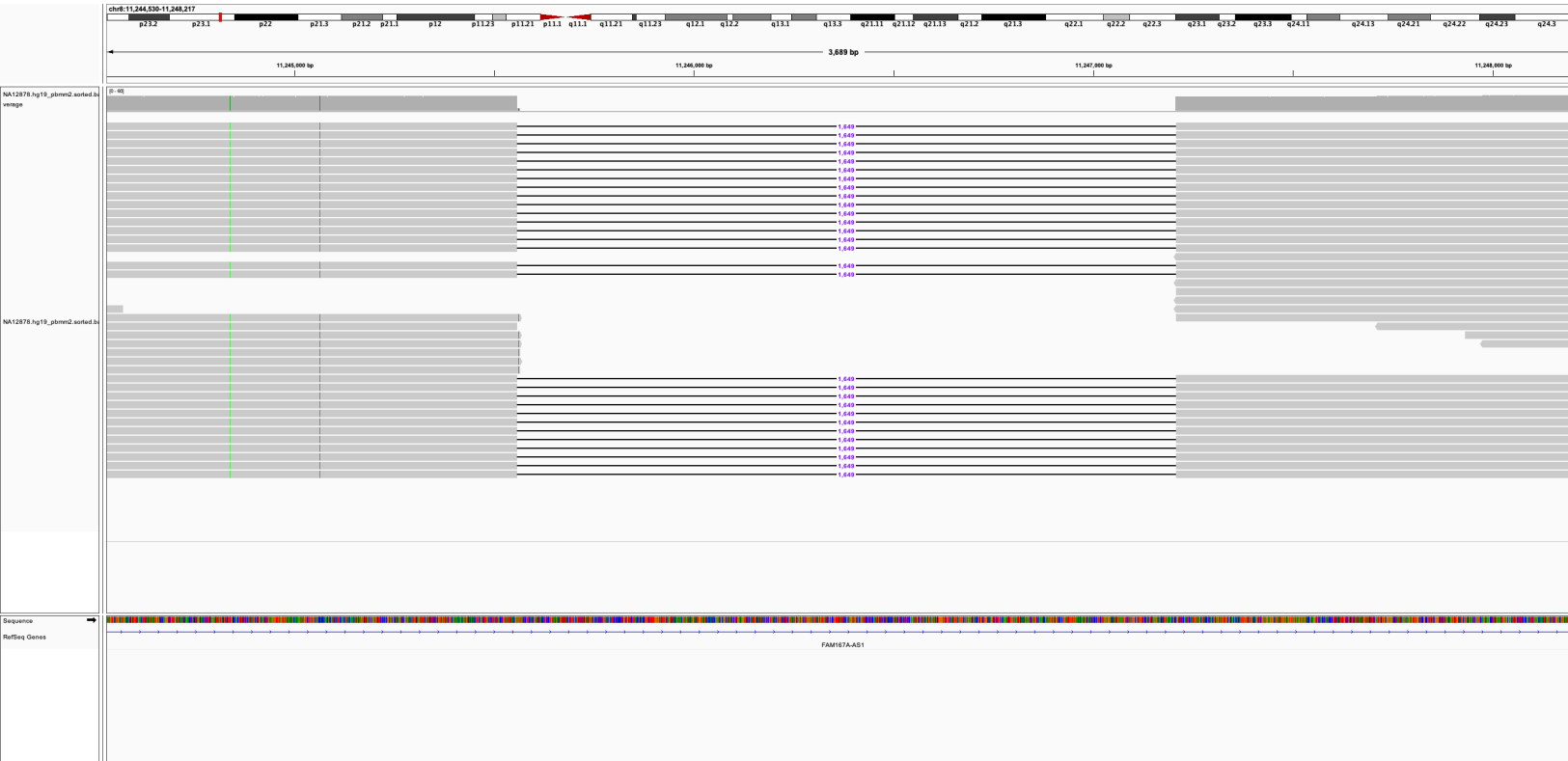
❖ chr7:81,441,546-81,442,495



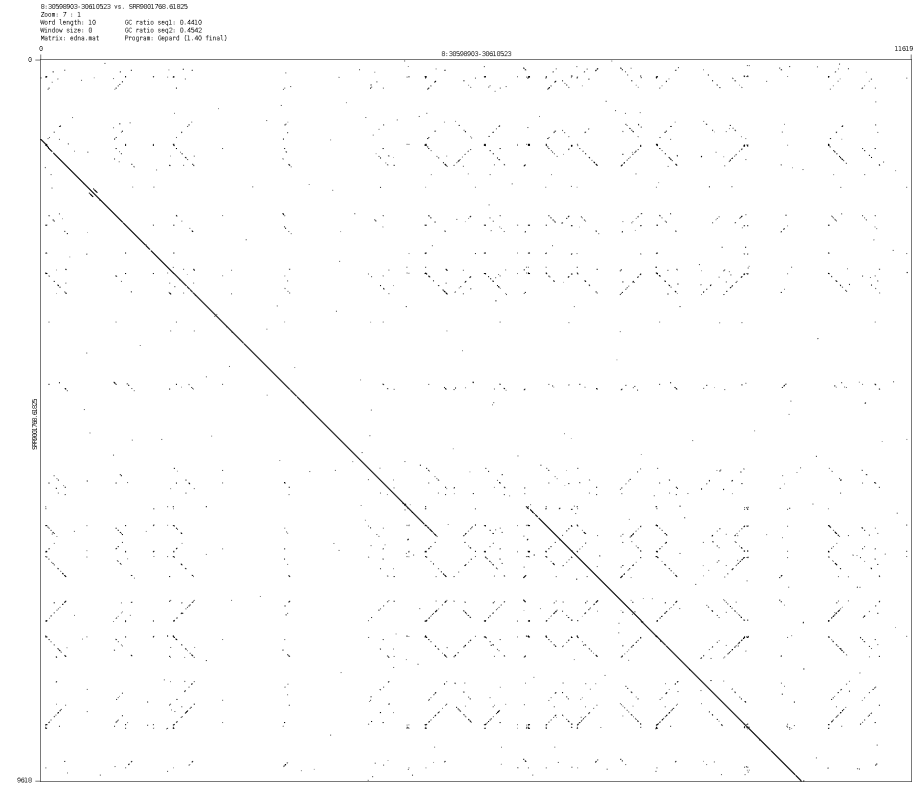
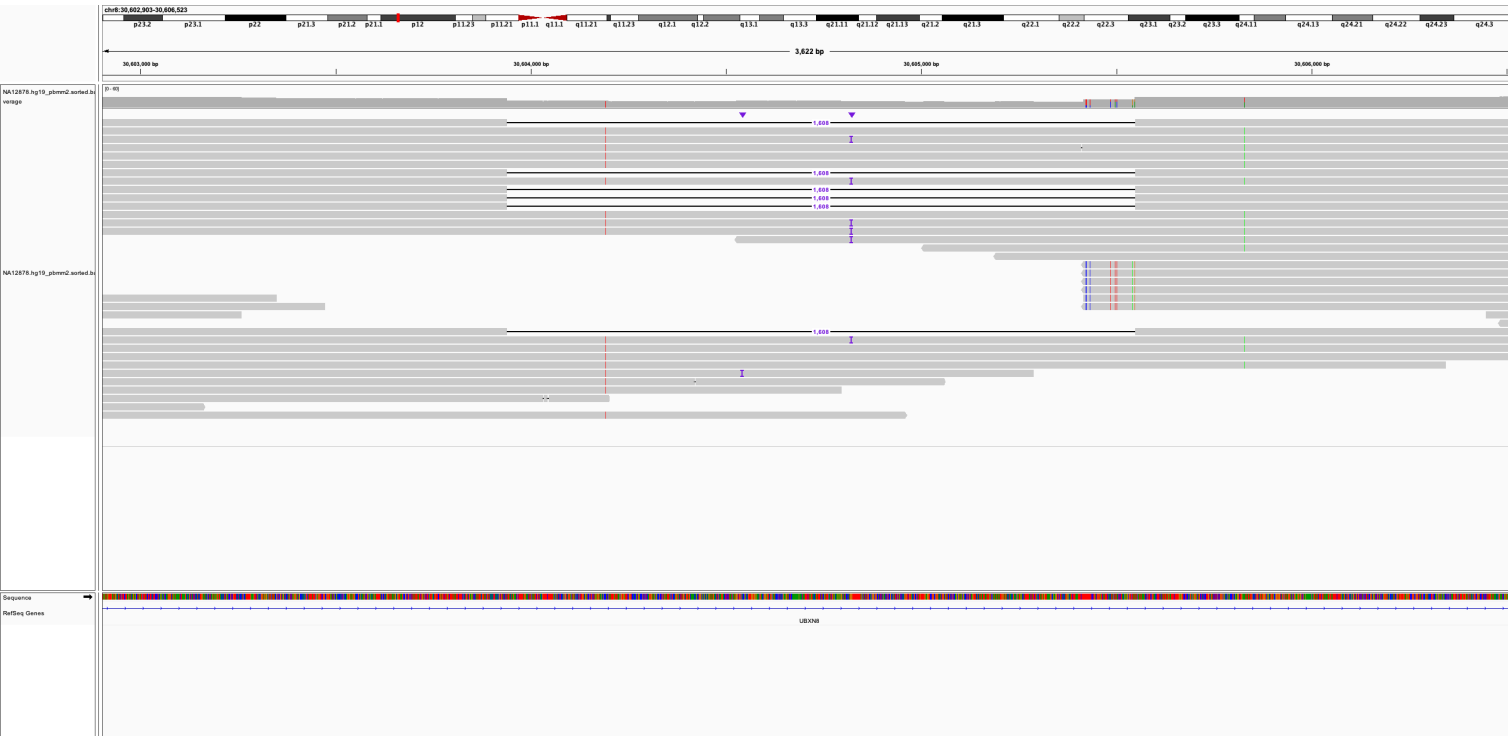
❖ chr7:138,322,188-138,322,252



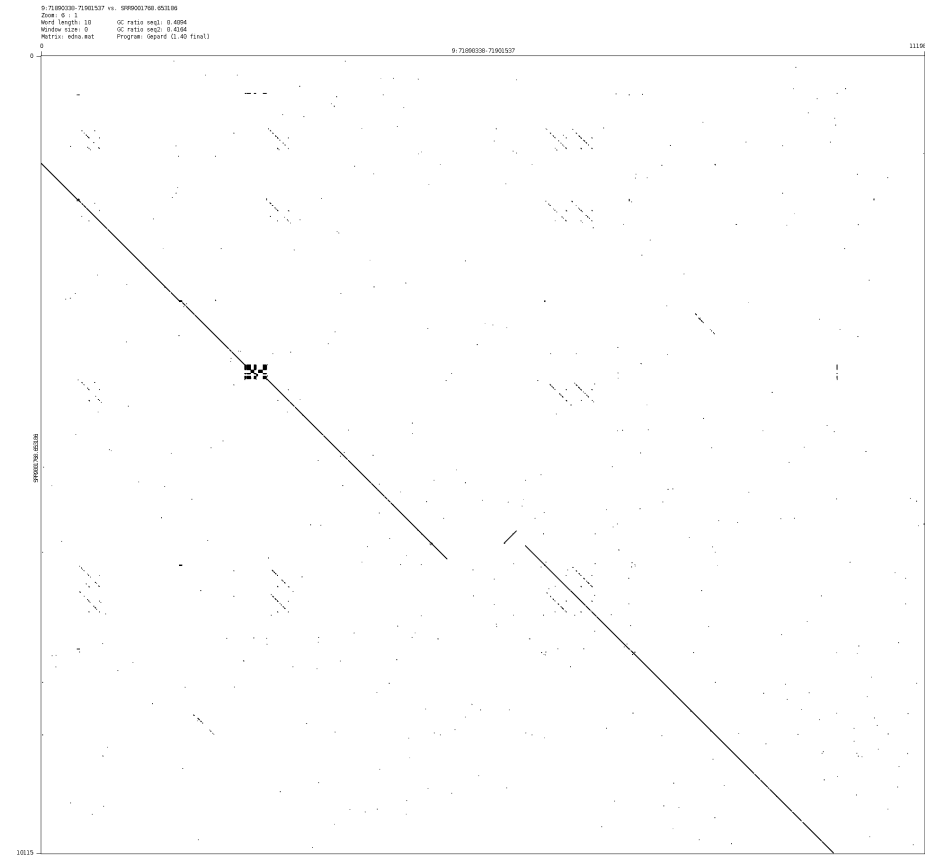
❖ chr8:11,245,530-11,247,217



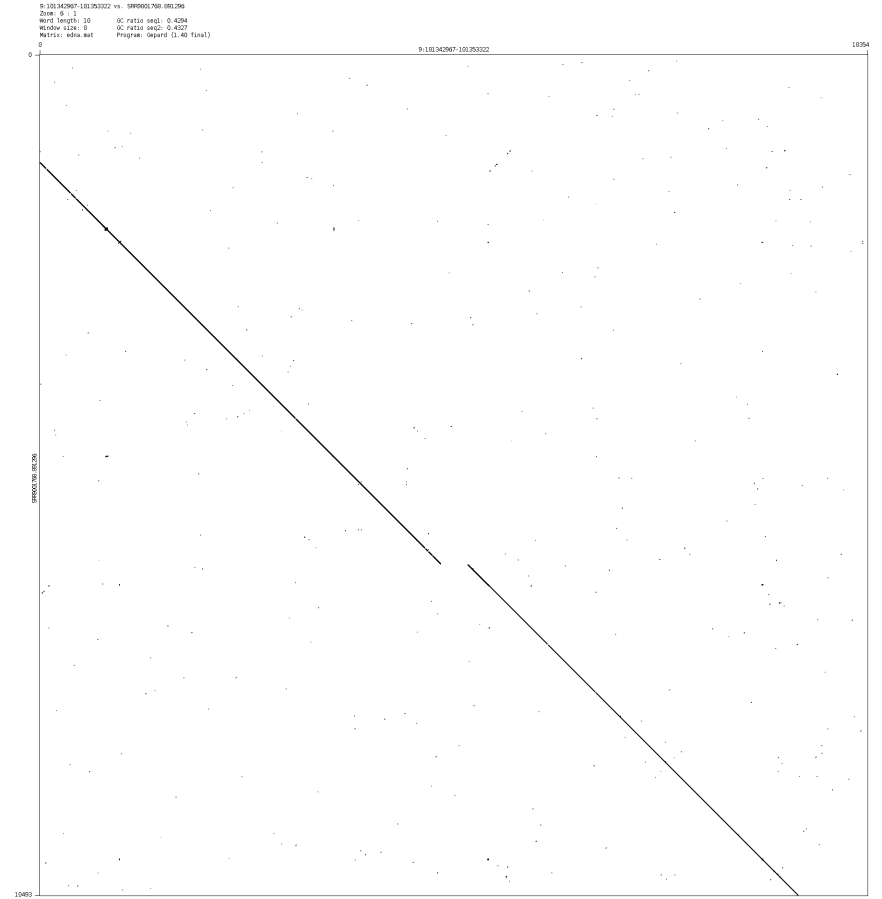
❖ chr8:30,603,903-30,605,523



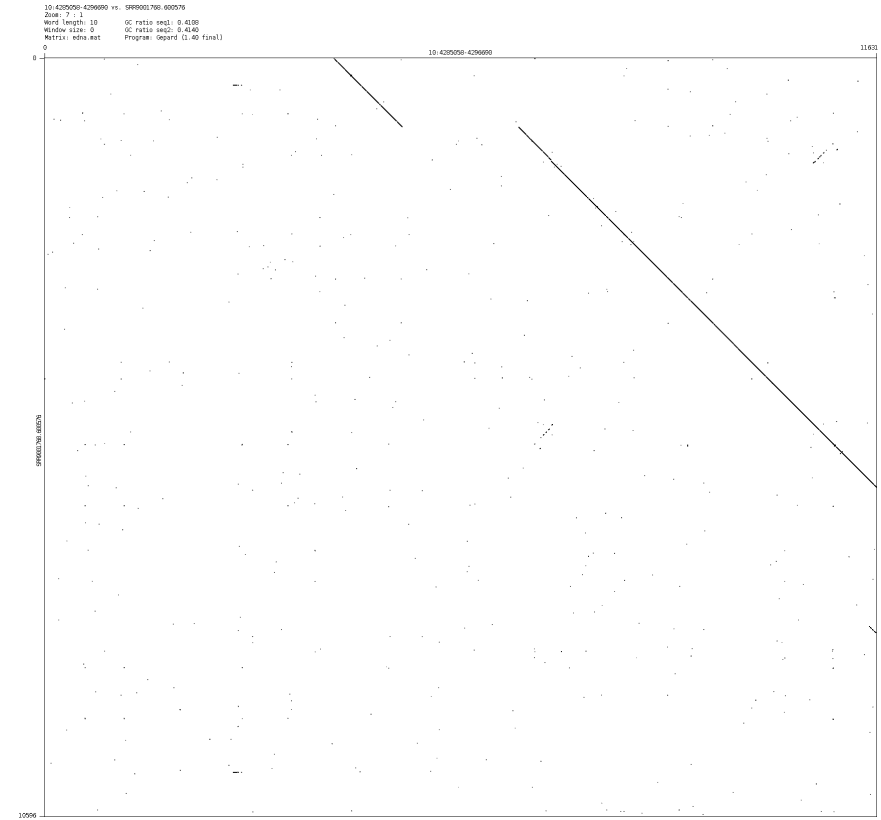
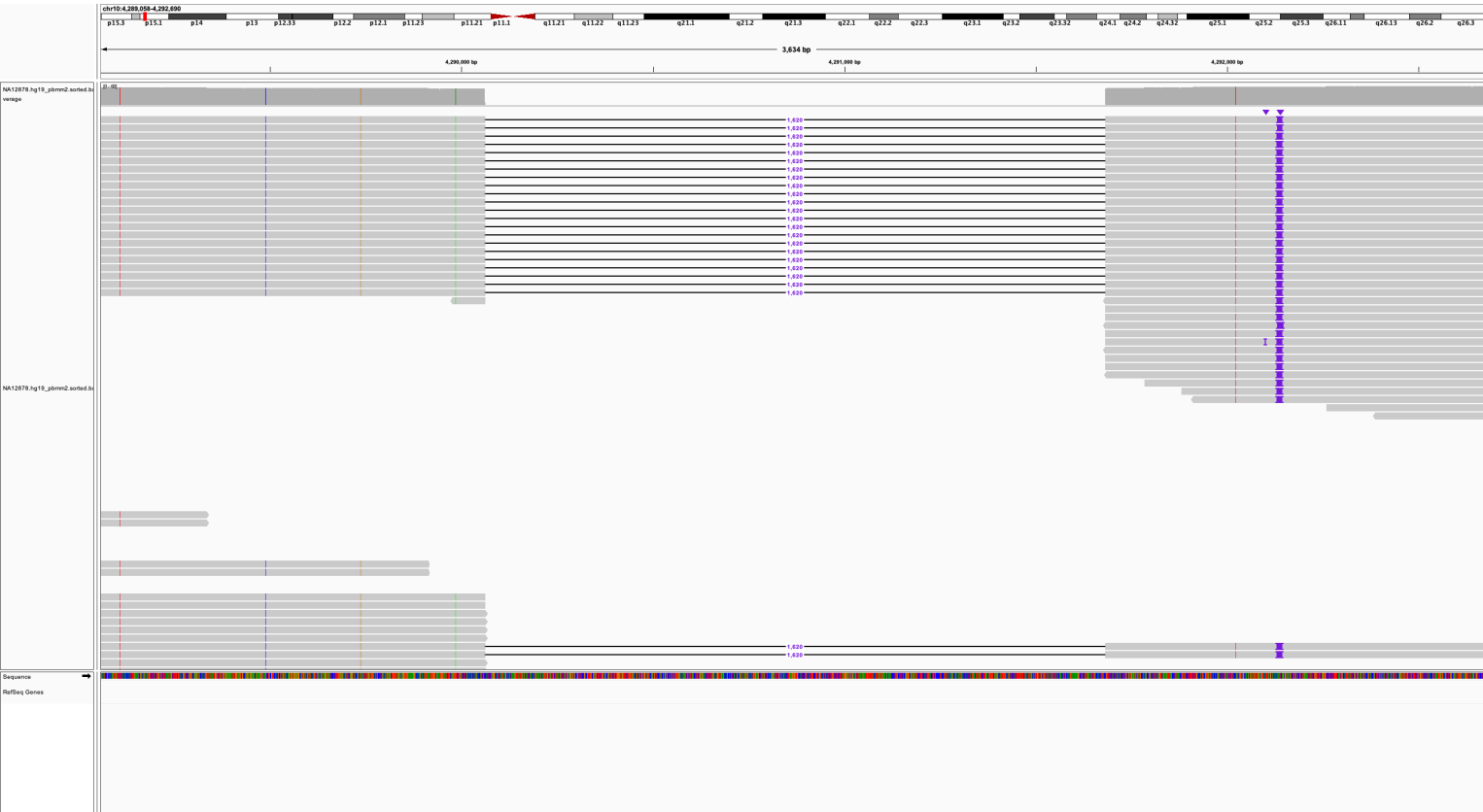
❖ chr9:71,895,338-71,896,537



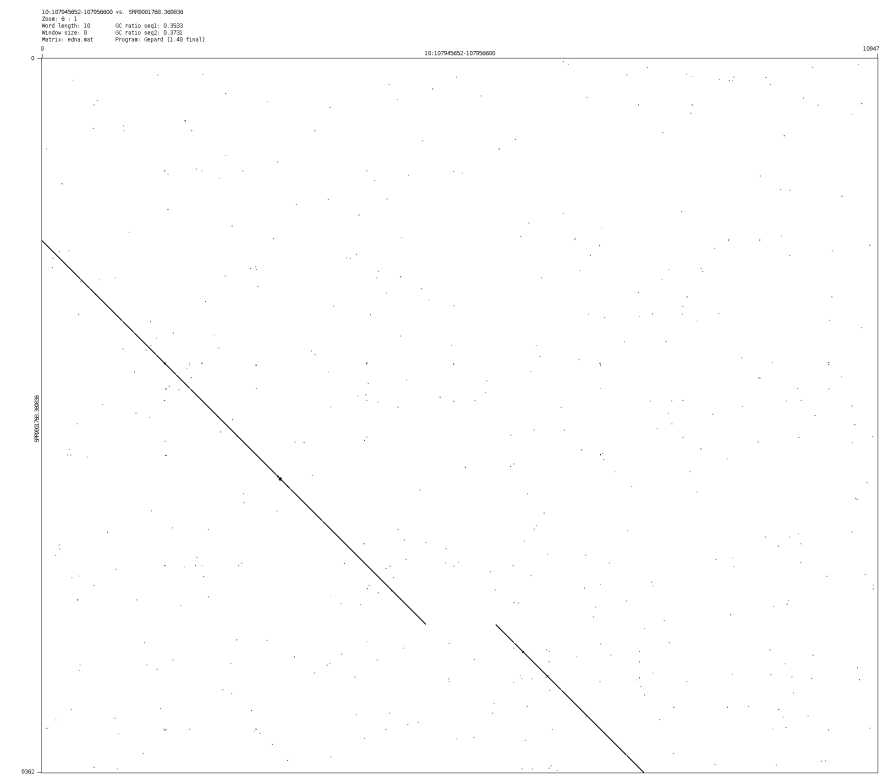
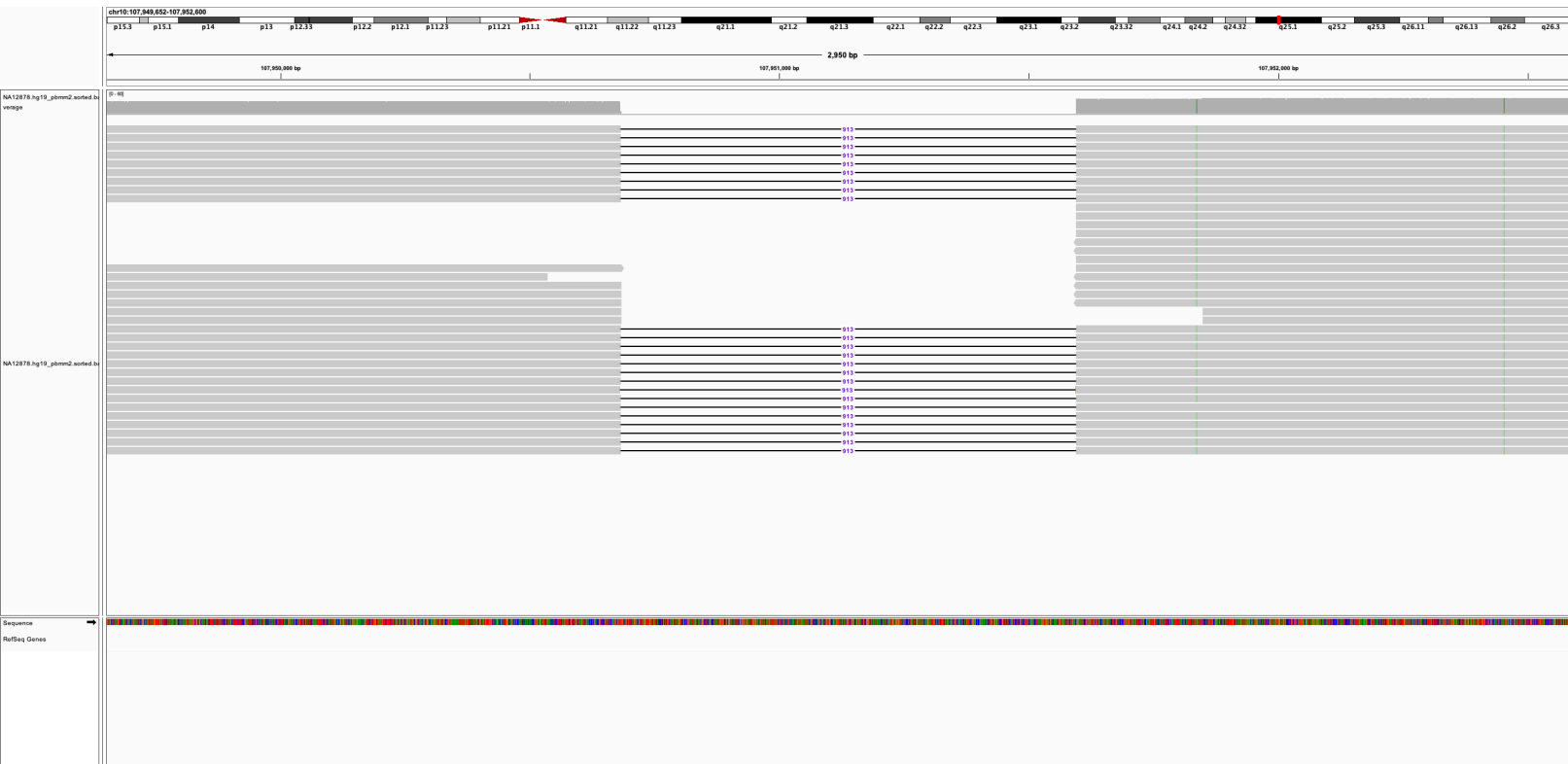
❖ chr9:101,347,967-101,348,322



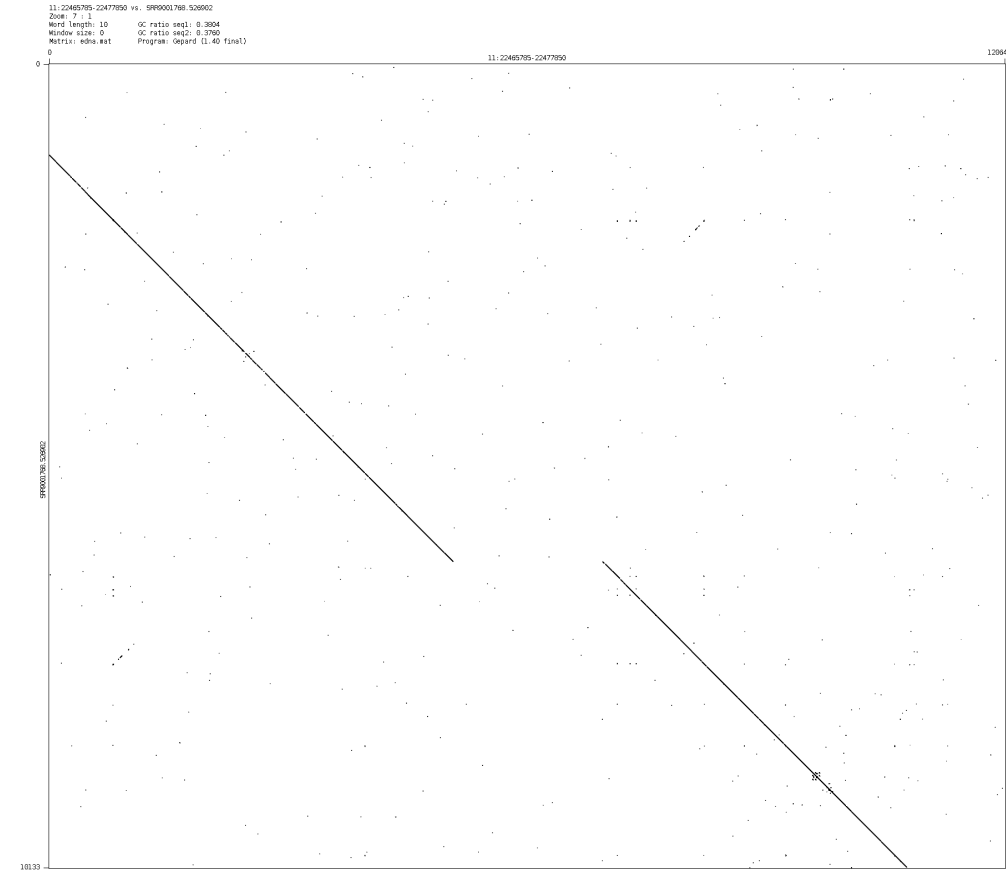
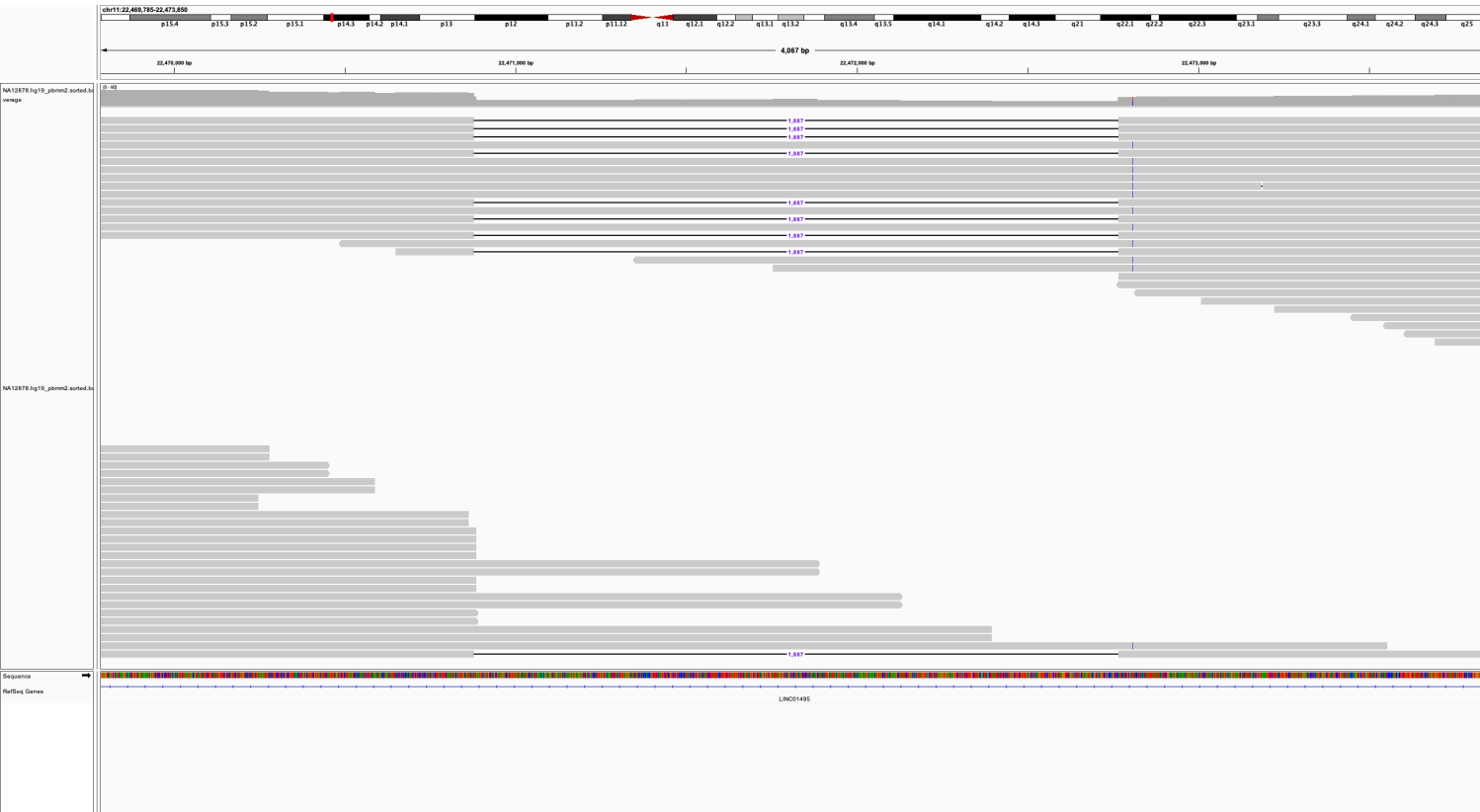
❖ chr10:4,290,058-4,291,690



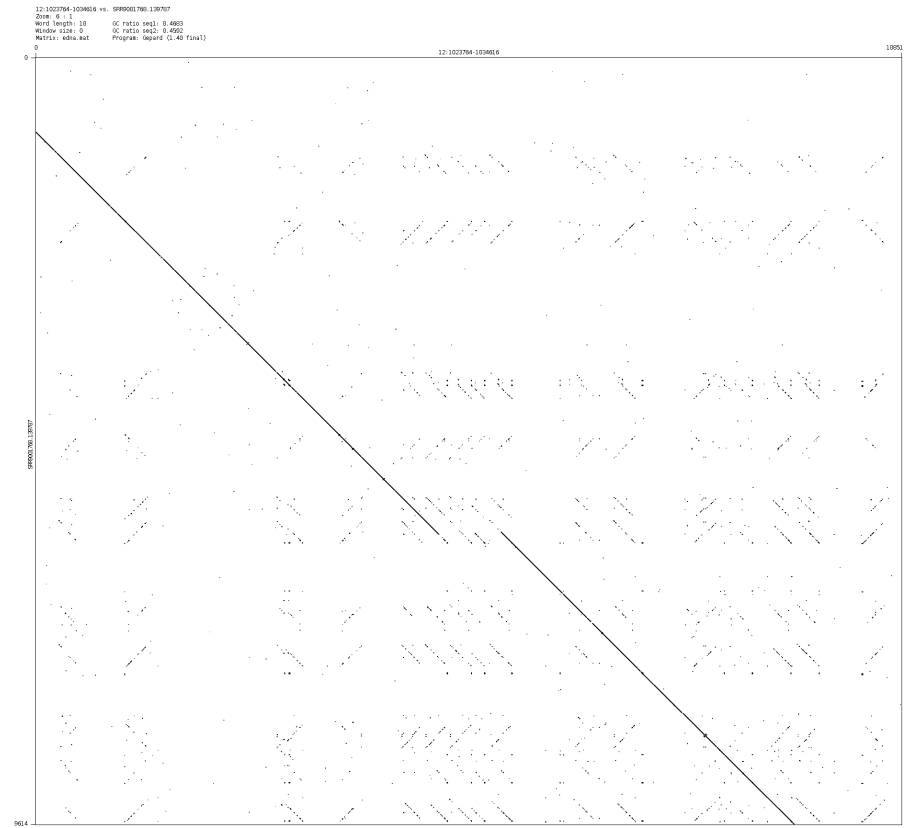
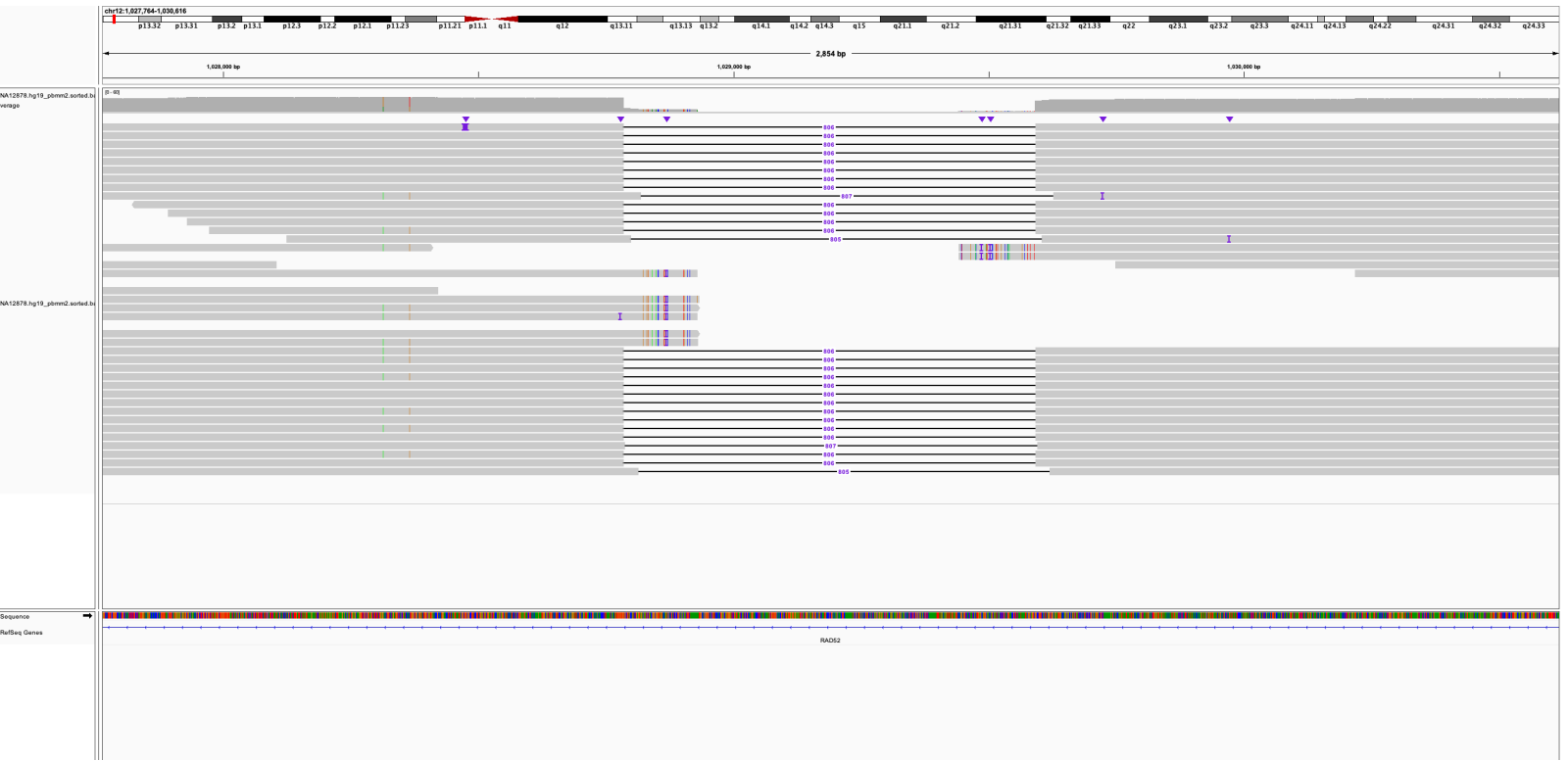
❖ chr10:107,950,652-107,951,600



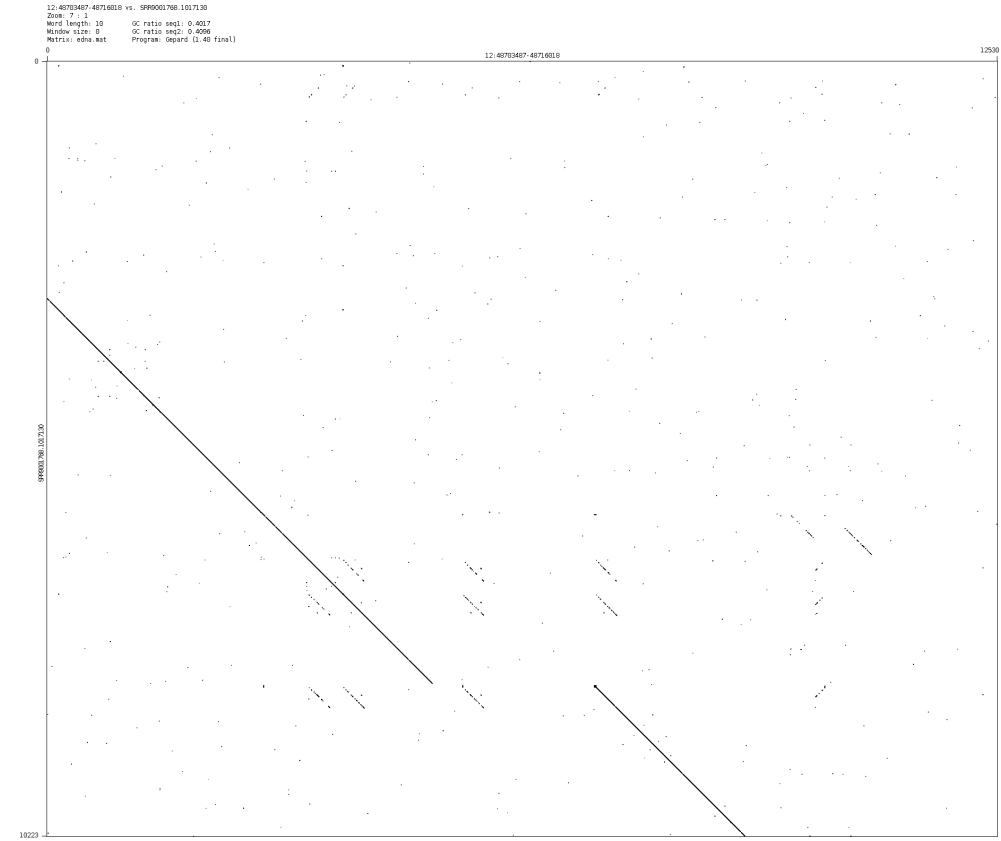
❖ chr11:22,470,785-22,472,850



❖ chr12:1,028,764-1,029,616

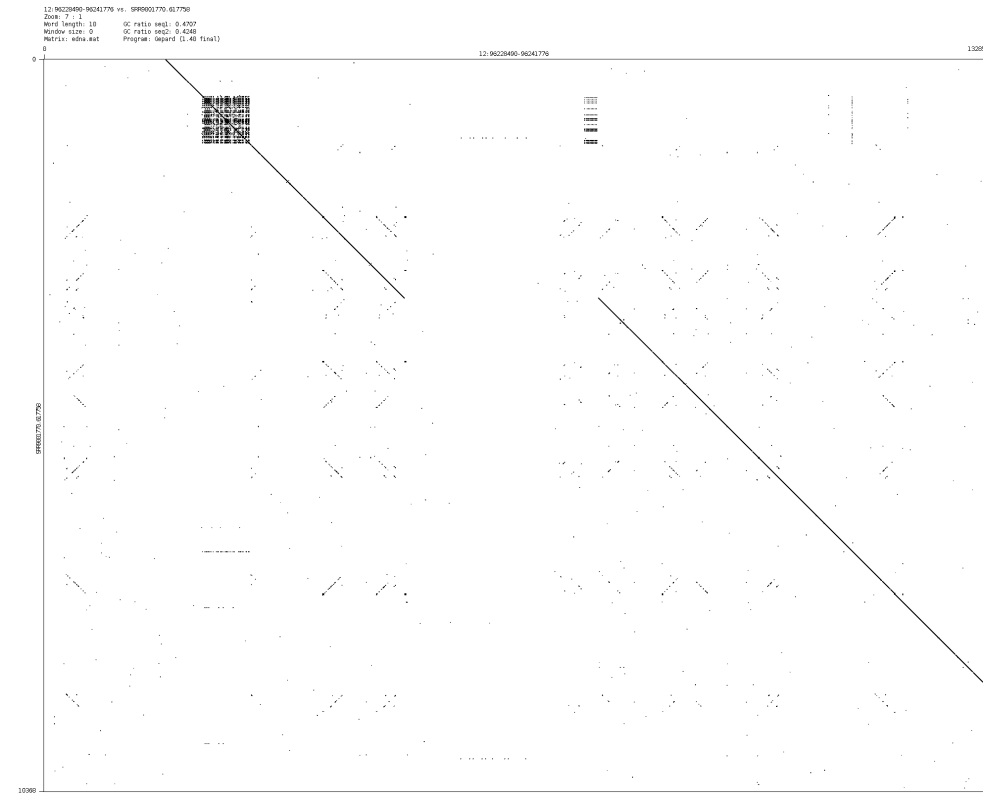
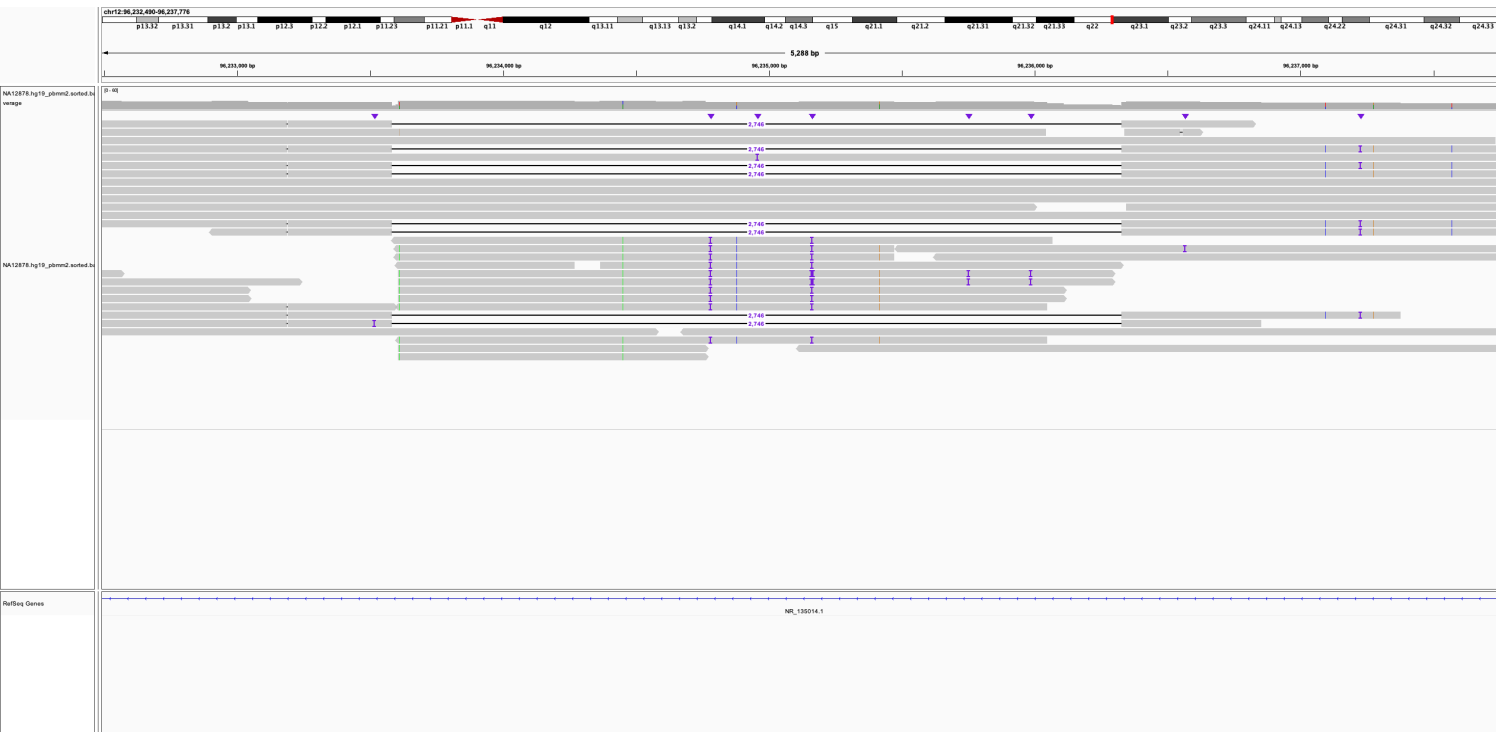


❖ chr12:48,708,487-48,711,018



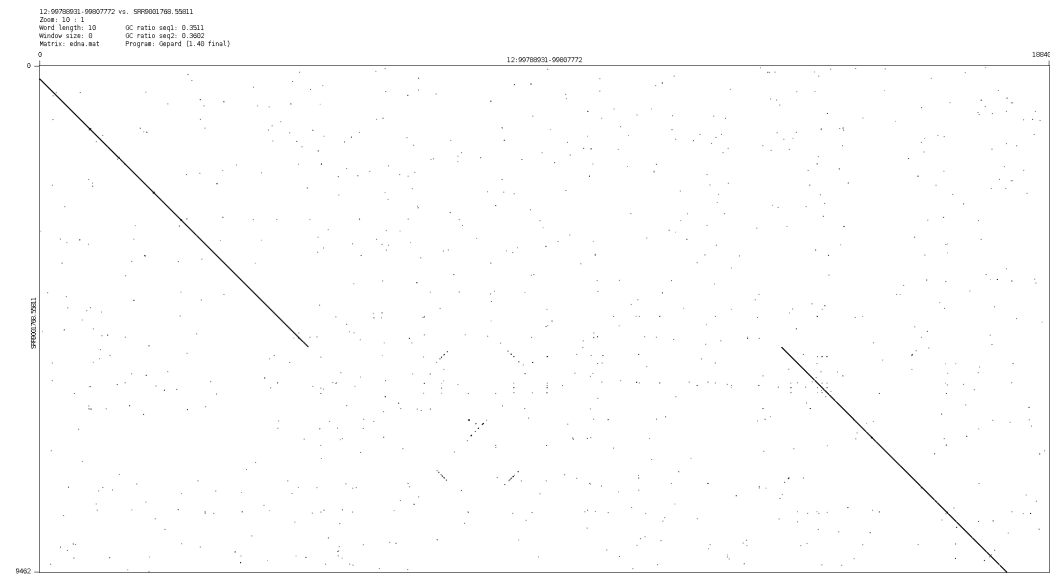
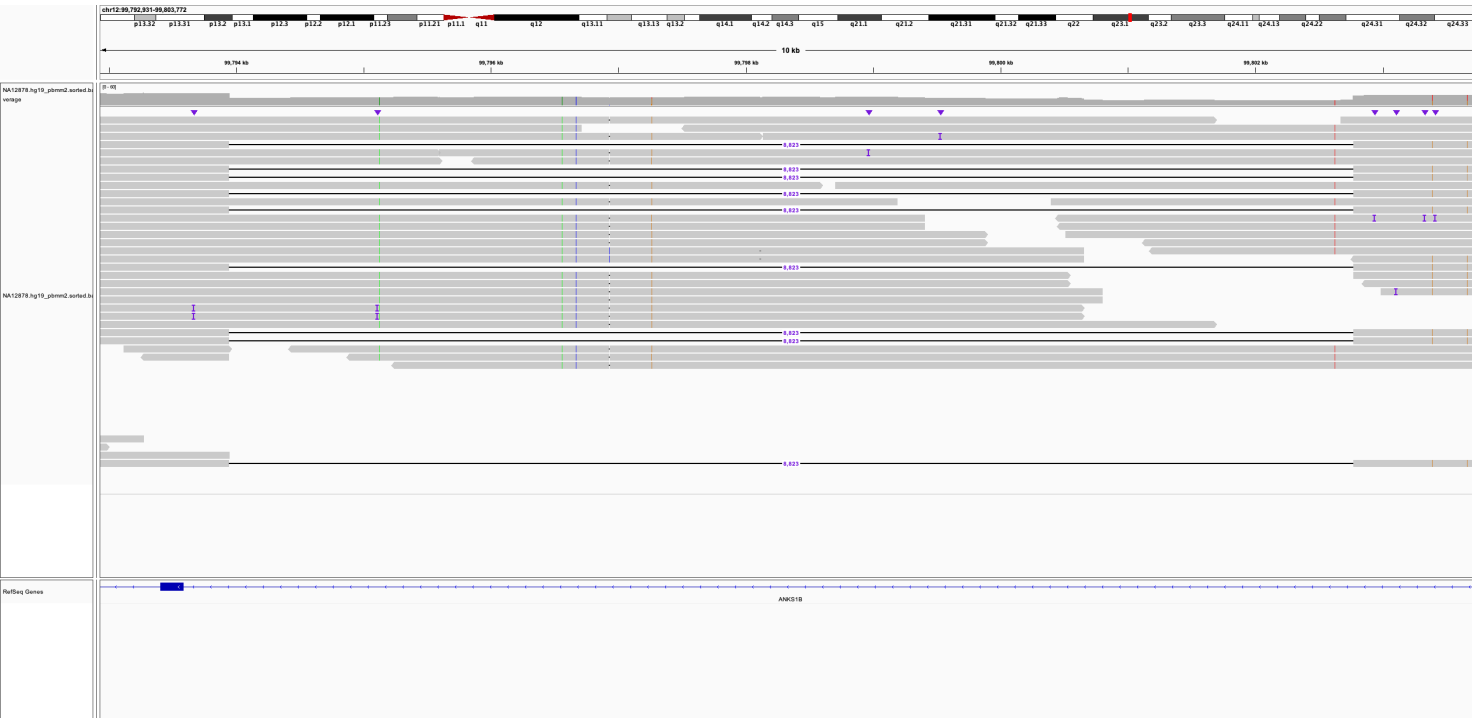


chr12:96,233,490-96,236,776



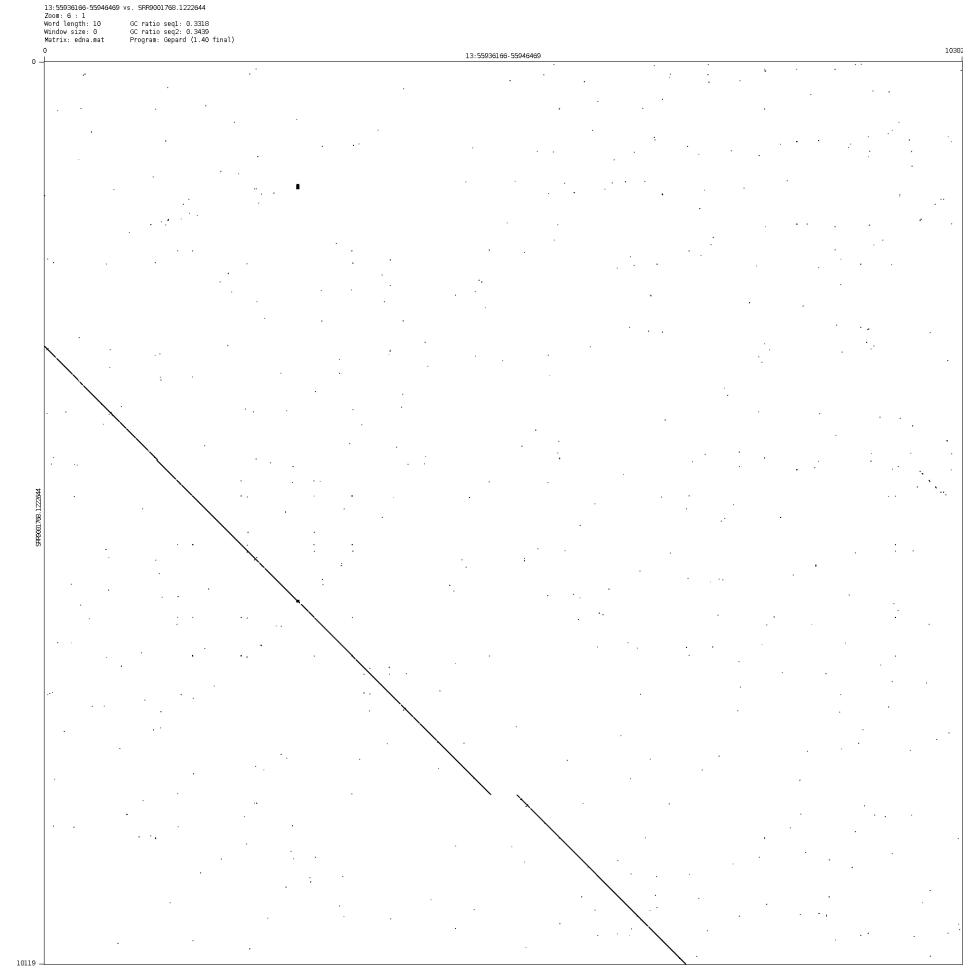


chr12:99,793,931-99,802,772





chr13:55,941,166-55,941,469



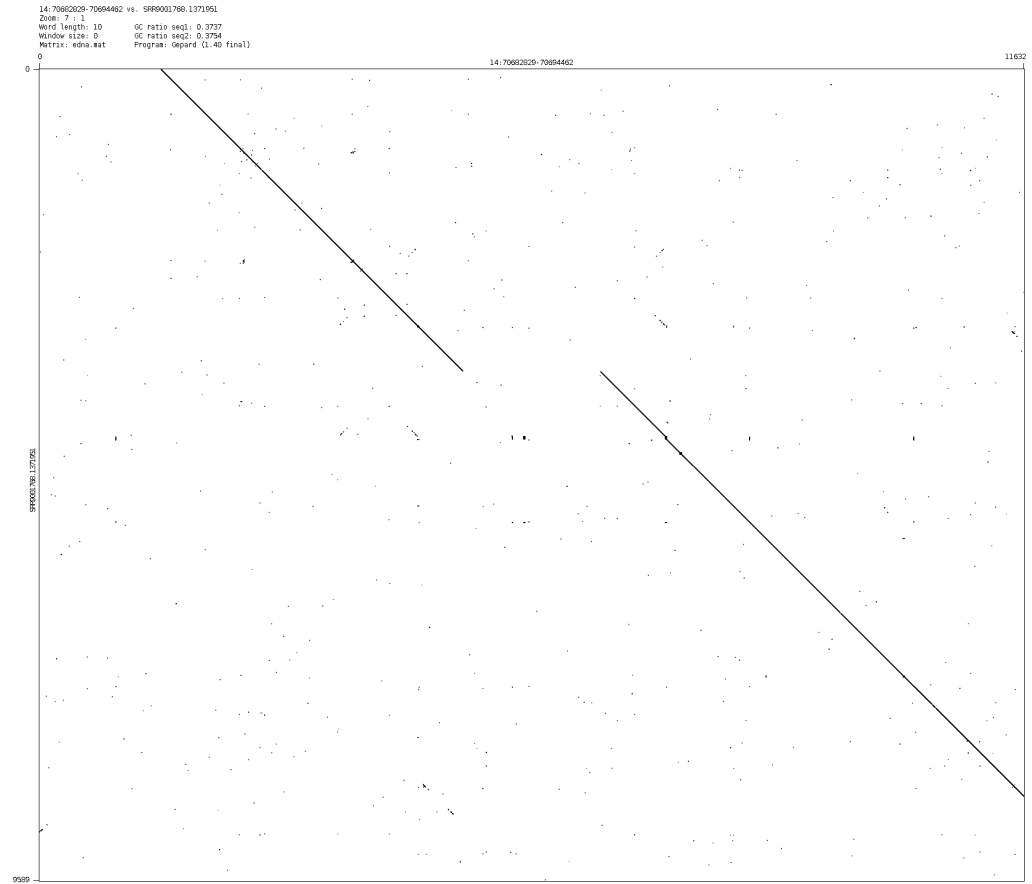
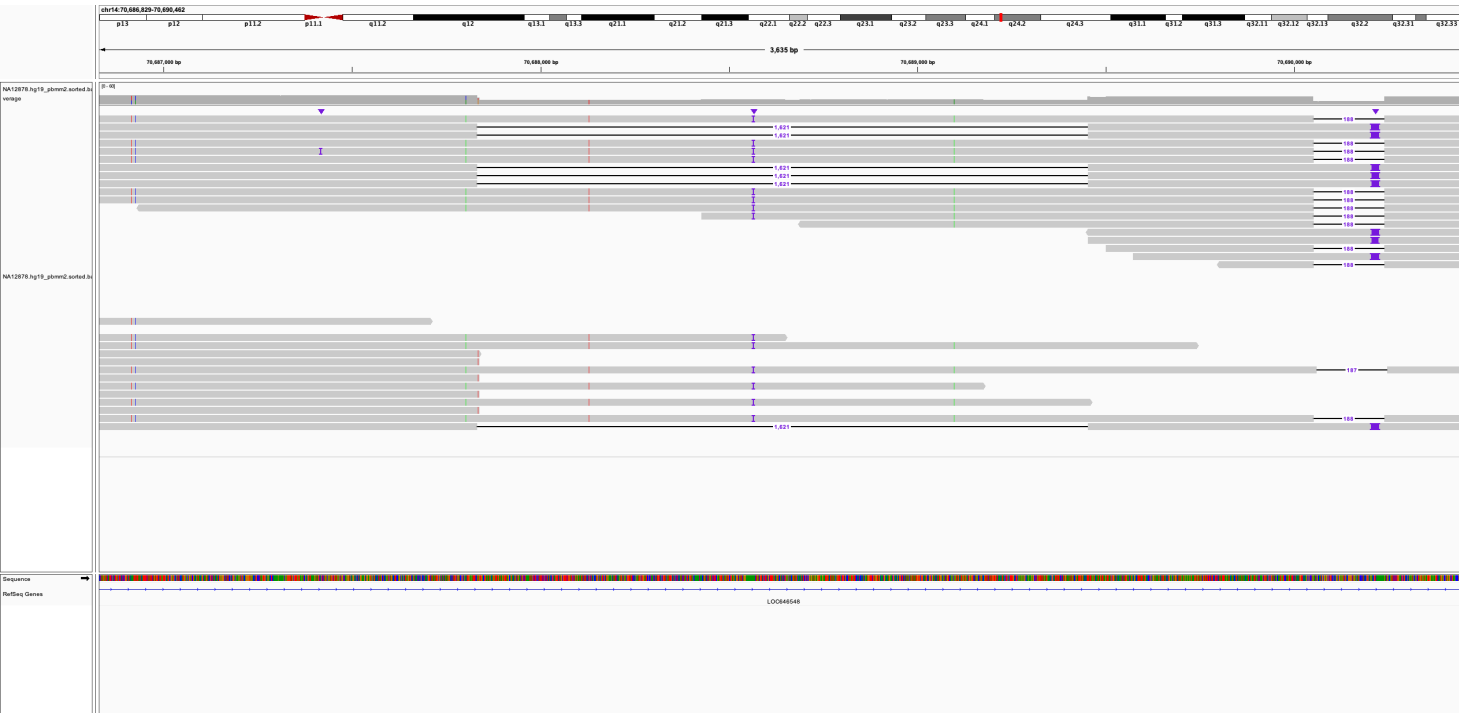


chr13:65,637,801-65,638,216



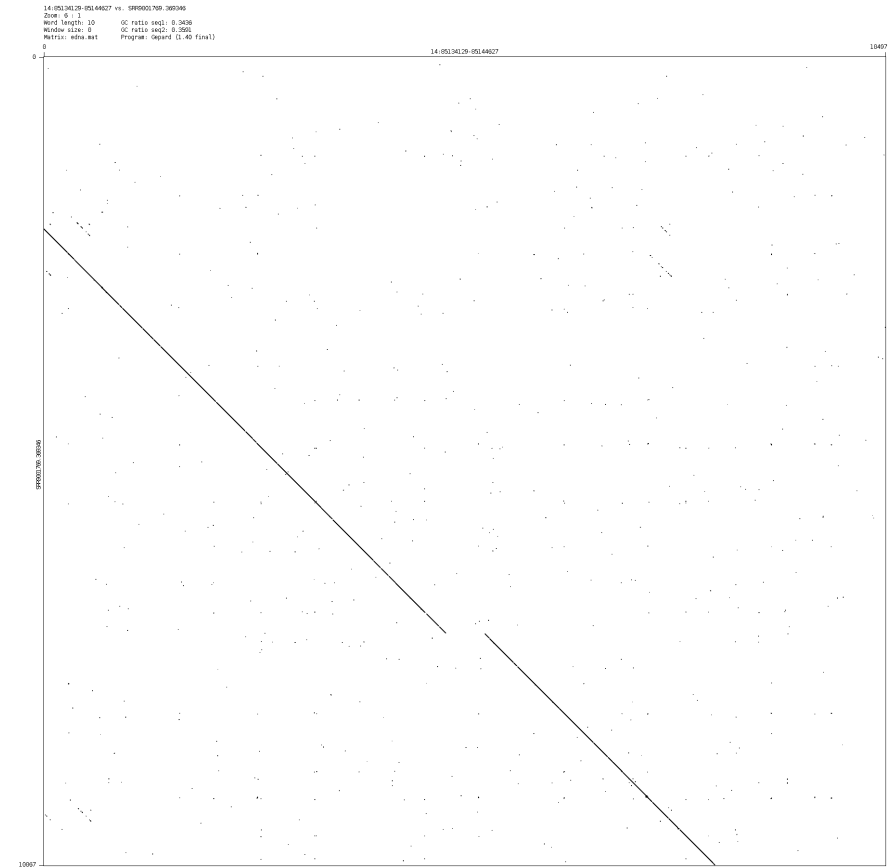
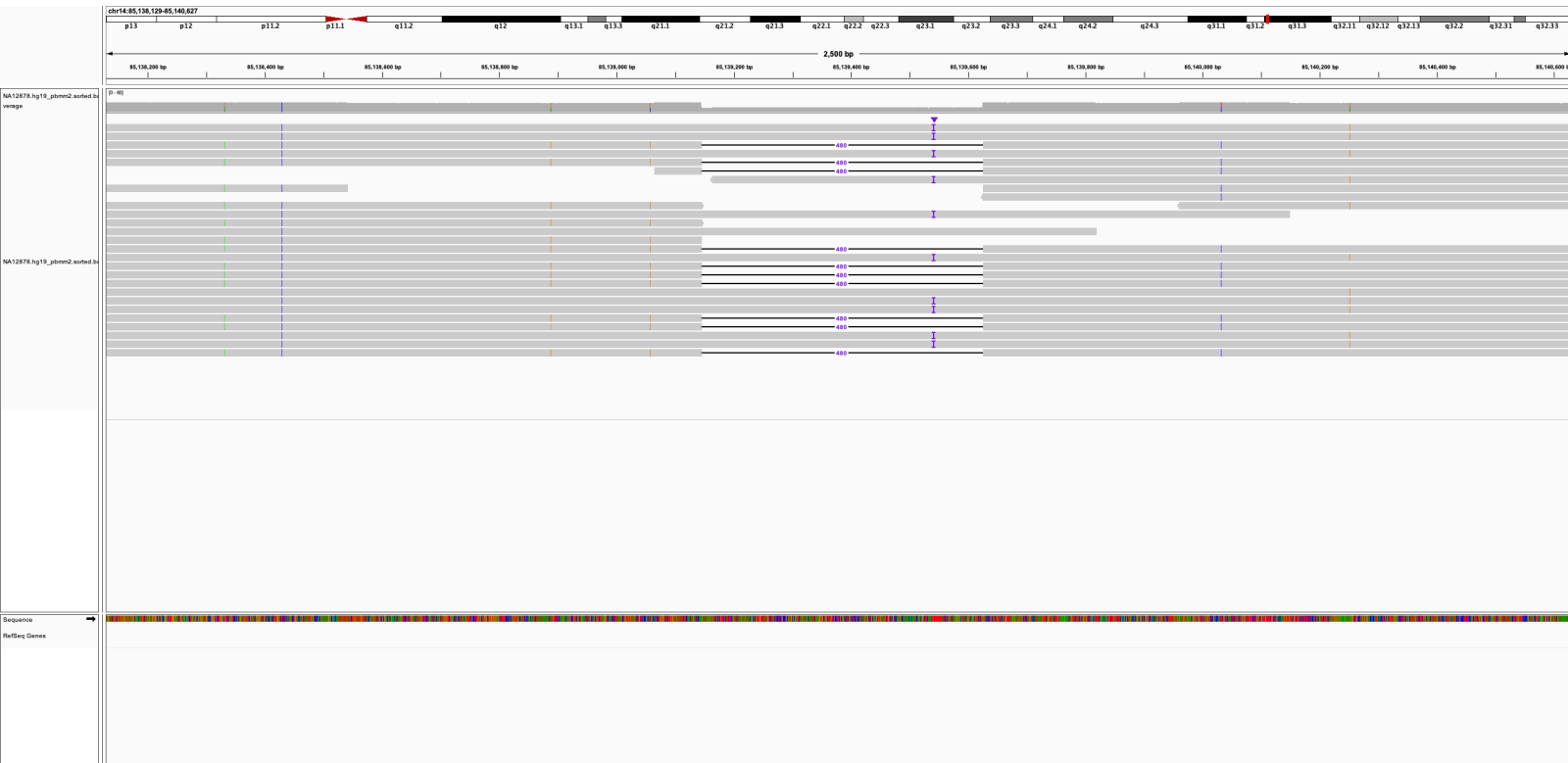


chr14:70,687,829-70,689,462

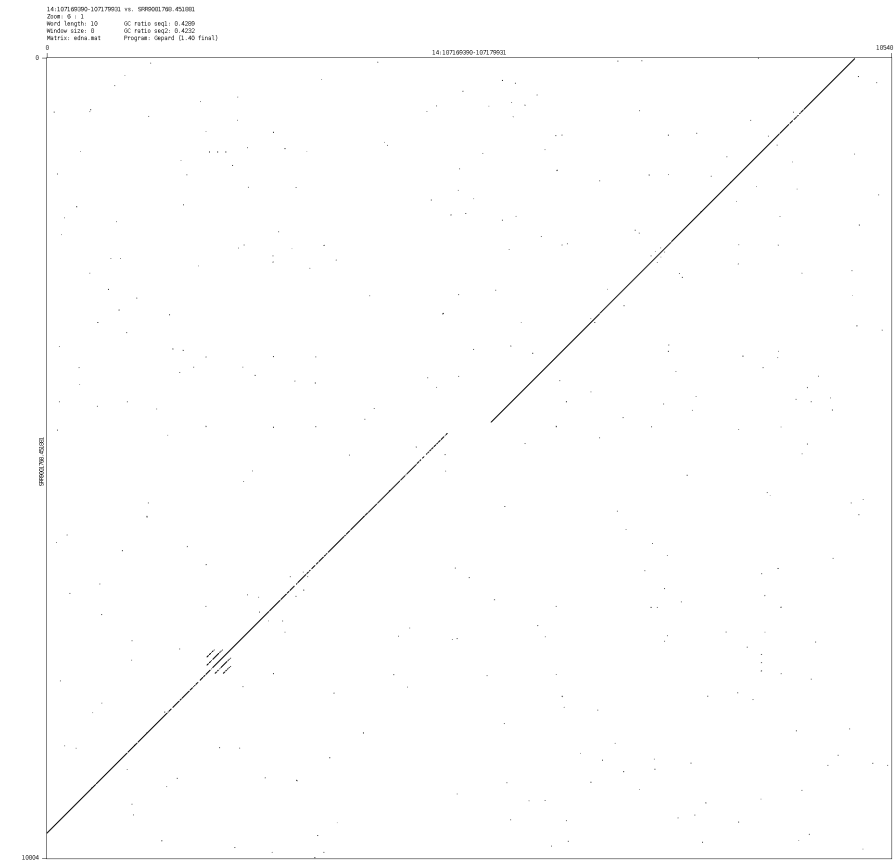




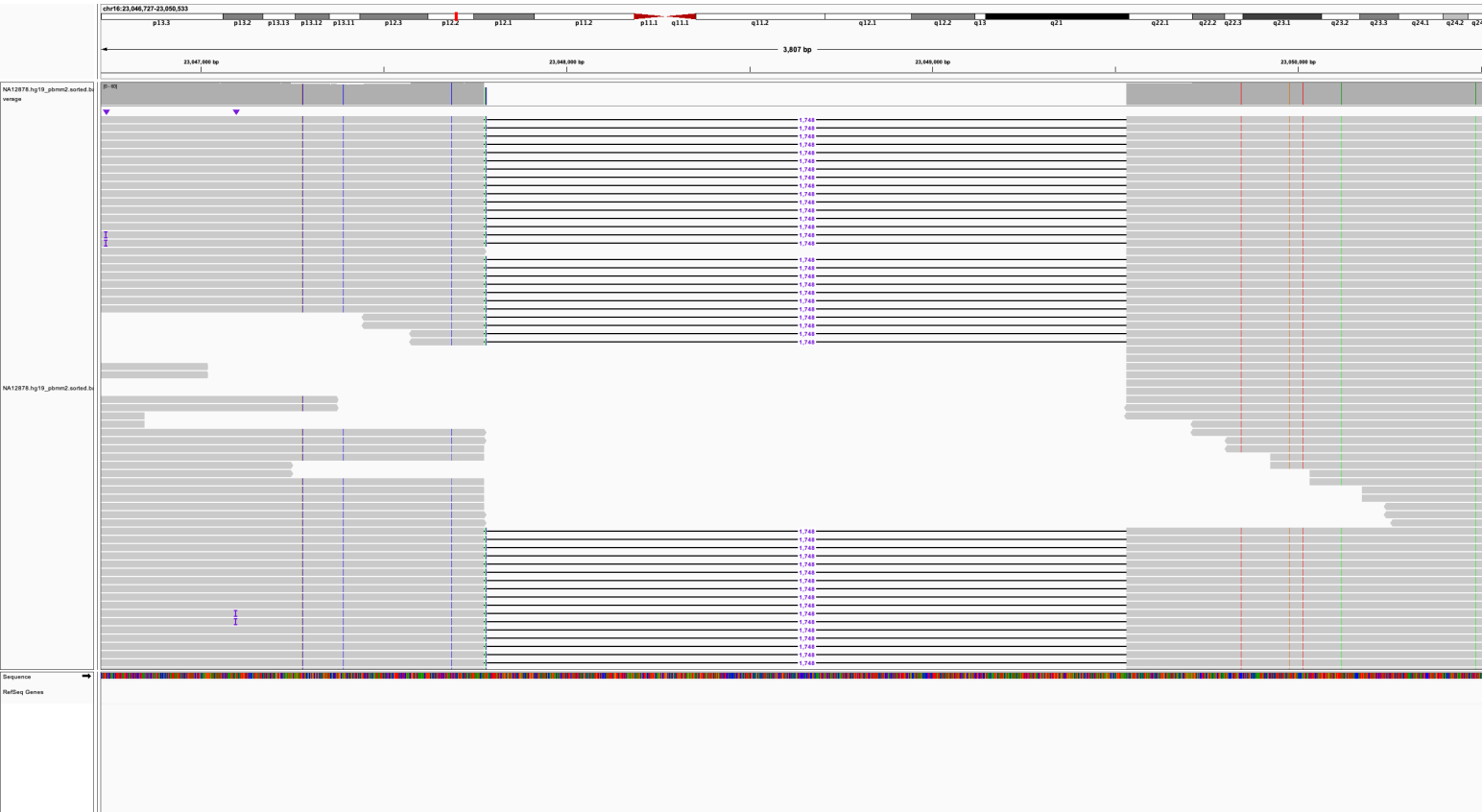
chr14:85,139,129-85,139,627



❖ chr14:107,174,390-107,174,931

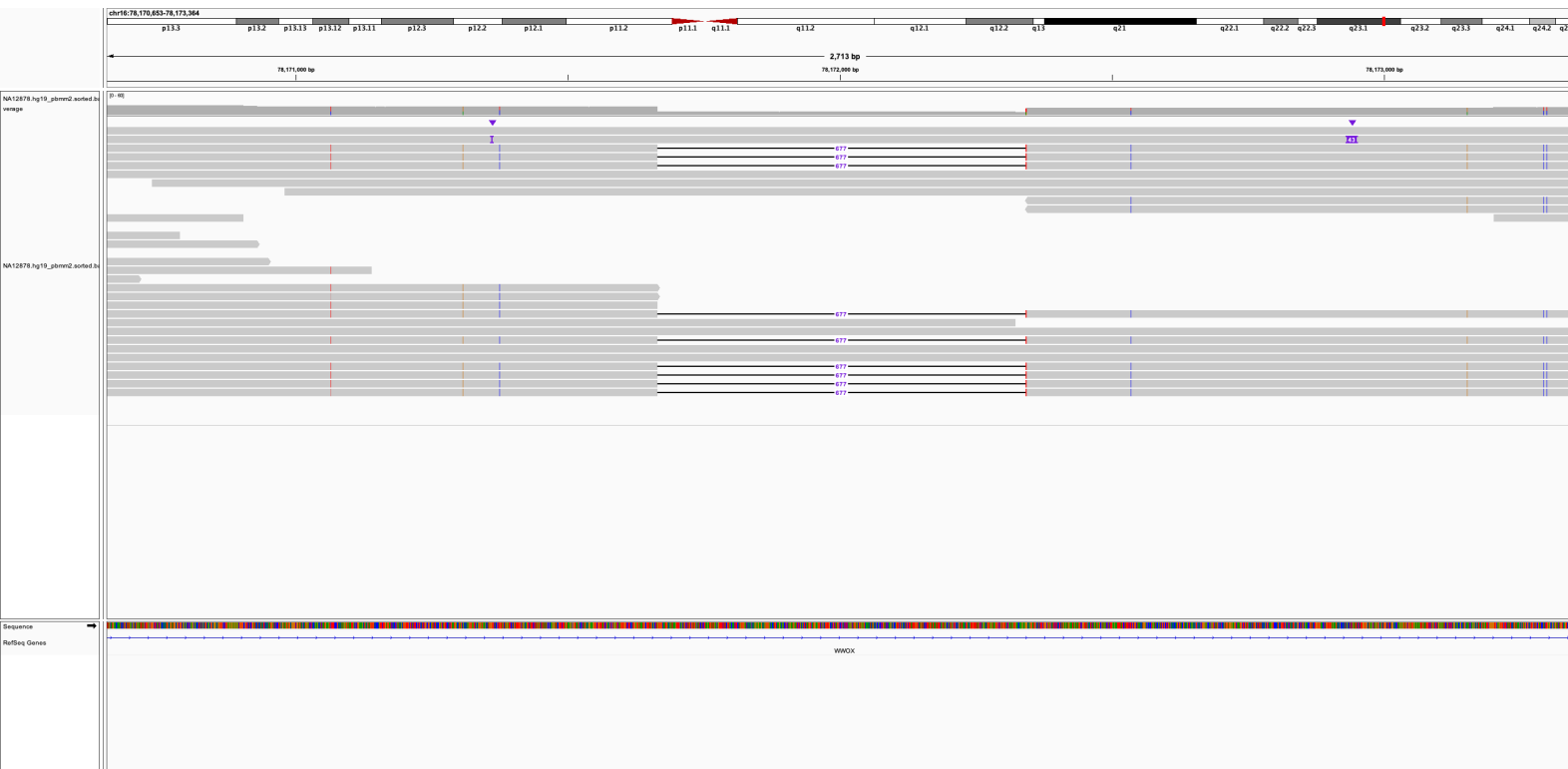


❖ chr16:23,047,727-23,049,533



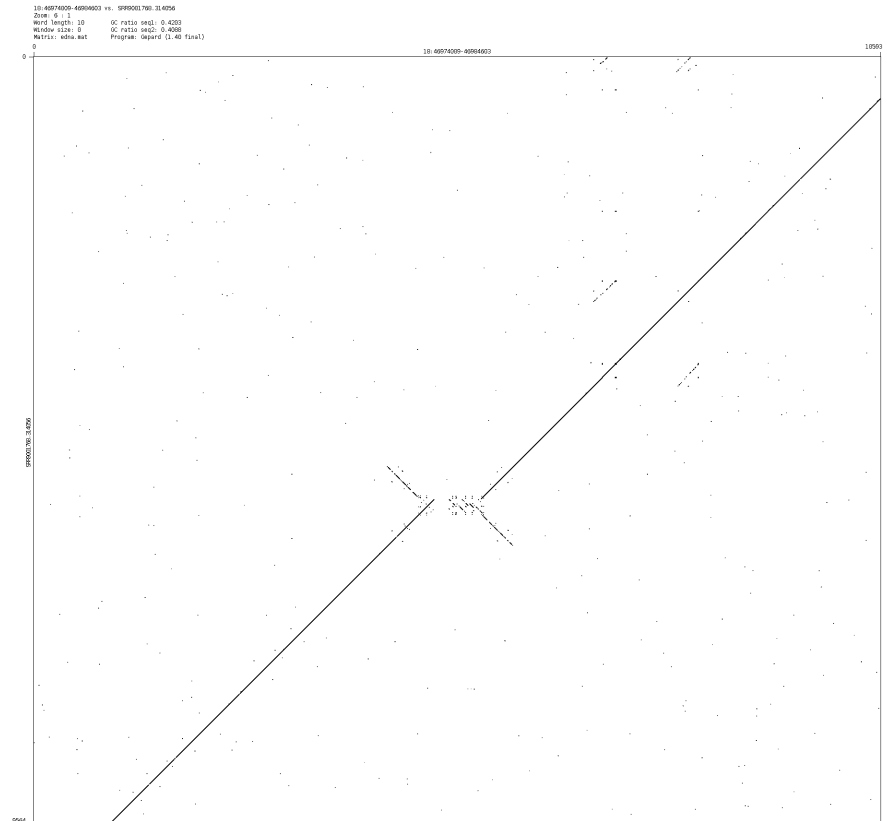
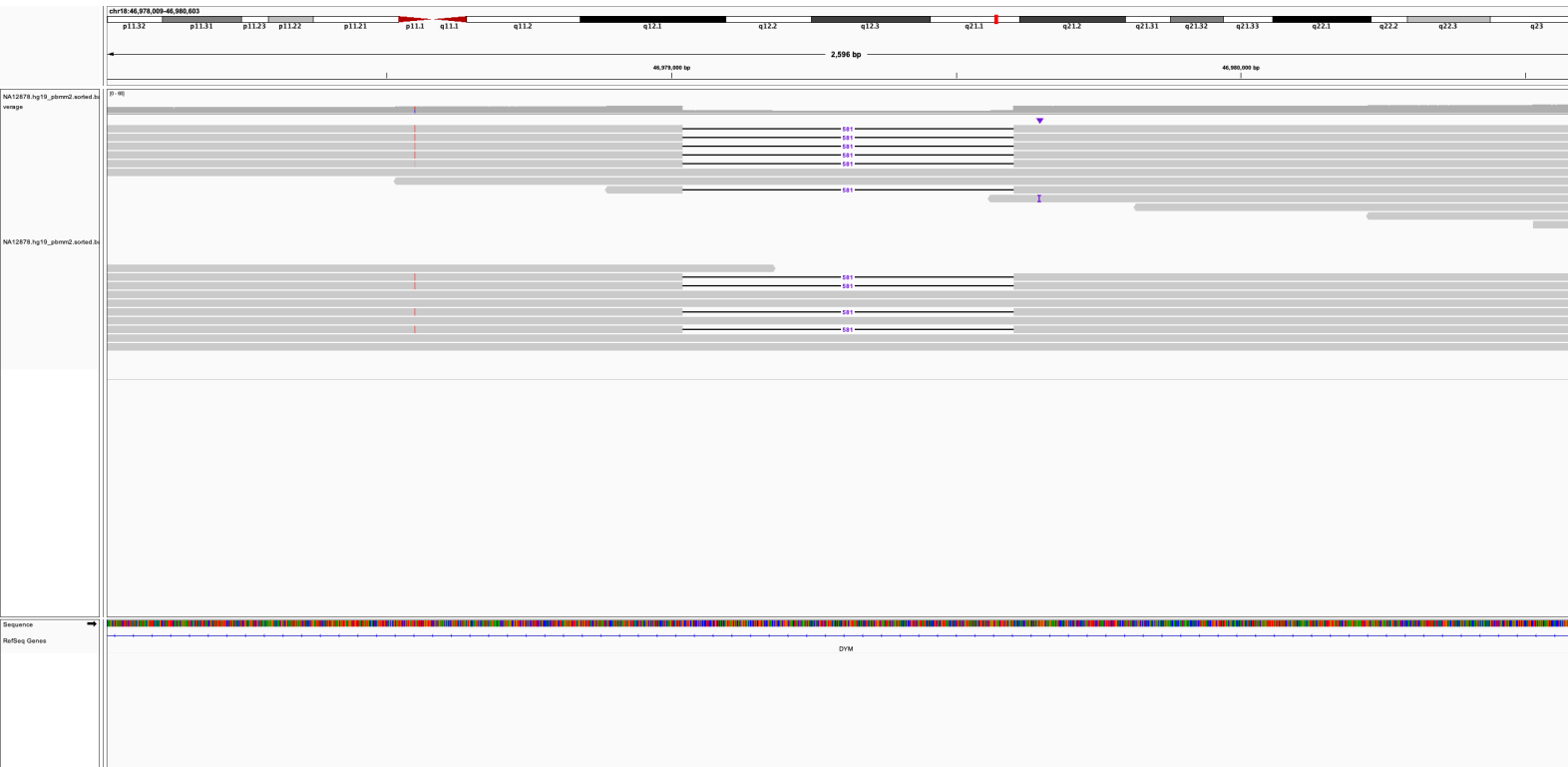


chr16:78,171,653-78,172,364

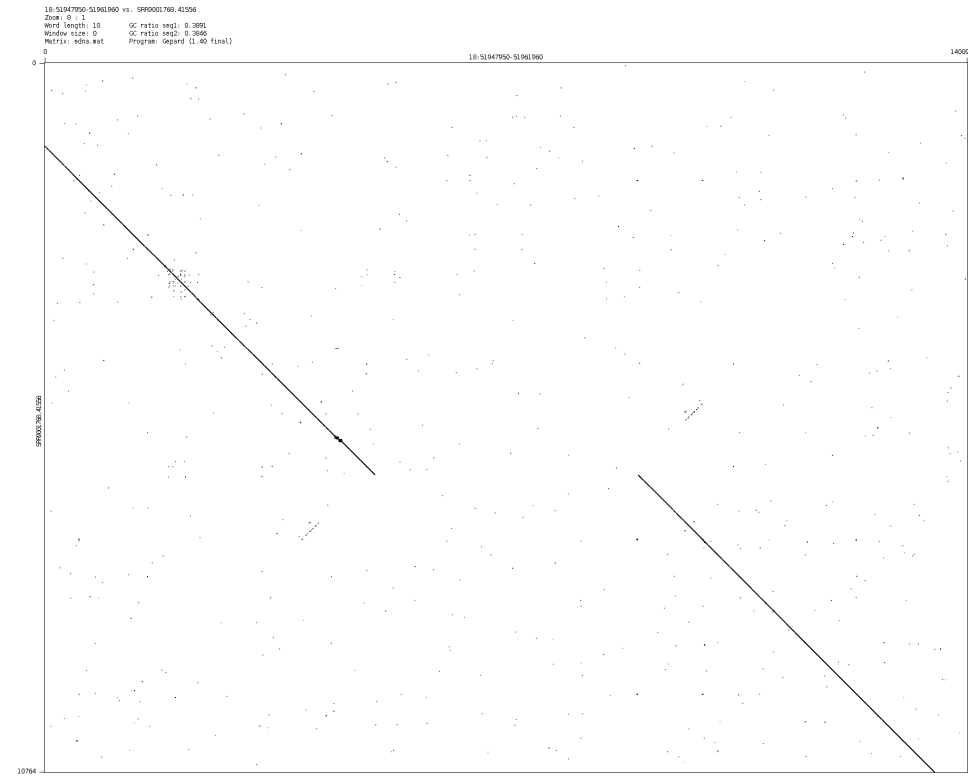




chr18:46,979,009-46,979,603

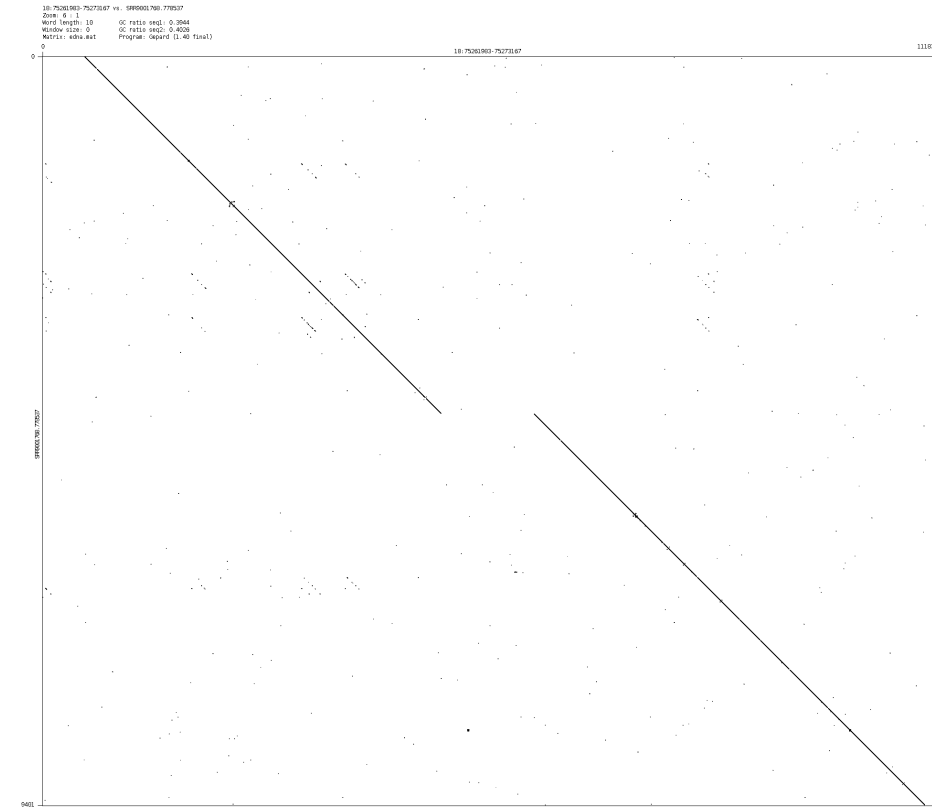
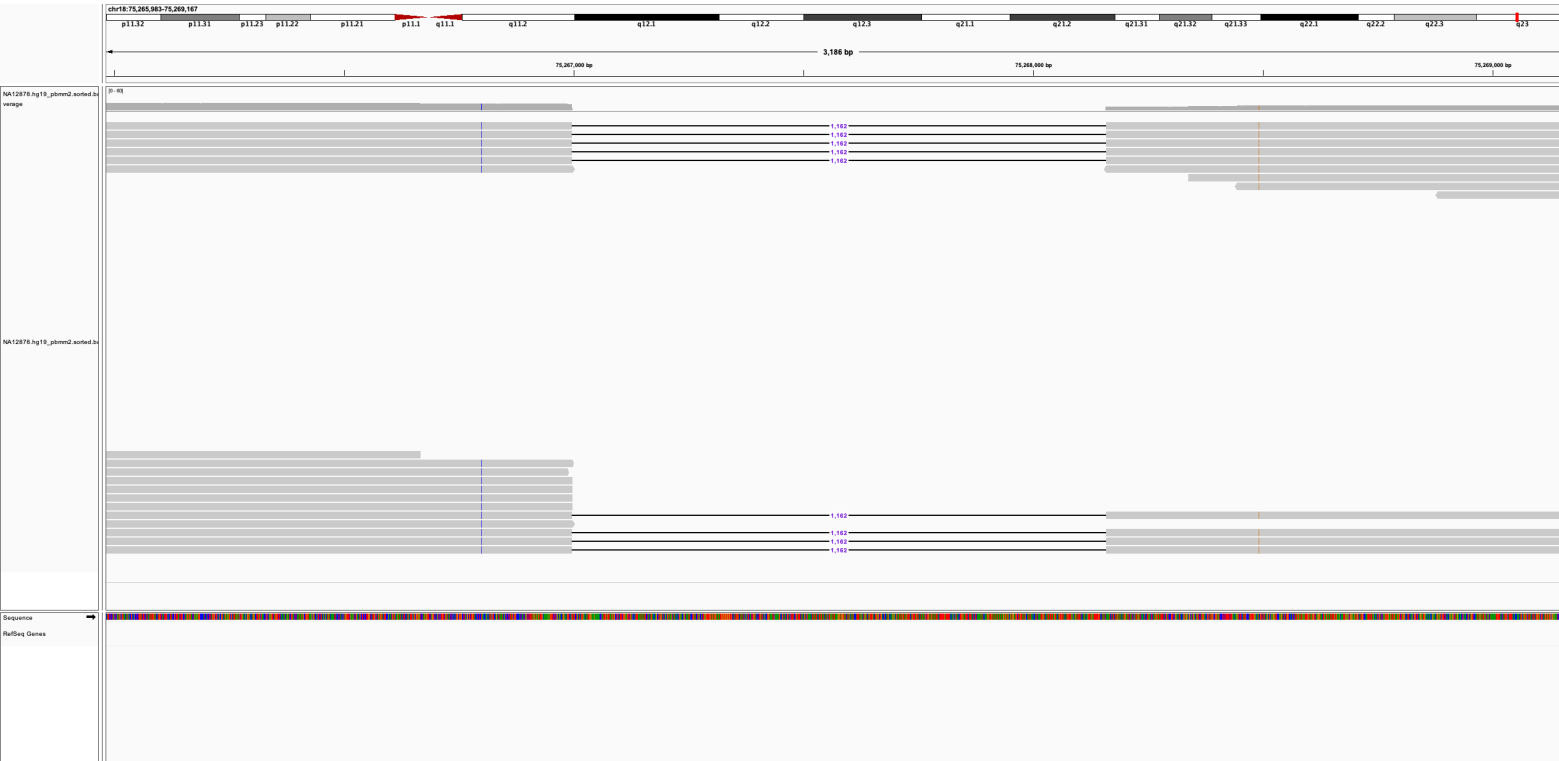


❖ chr18:51,952,950-51,956,960

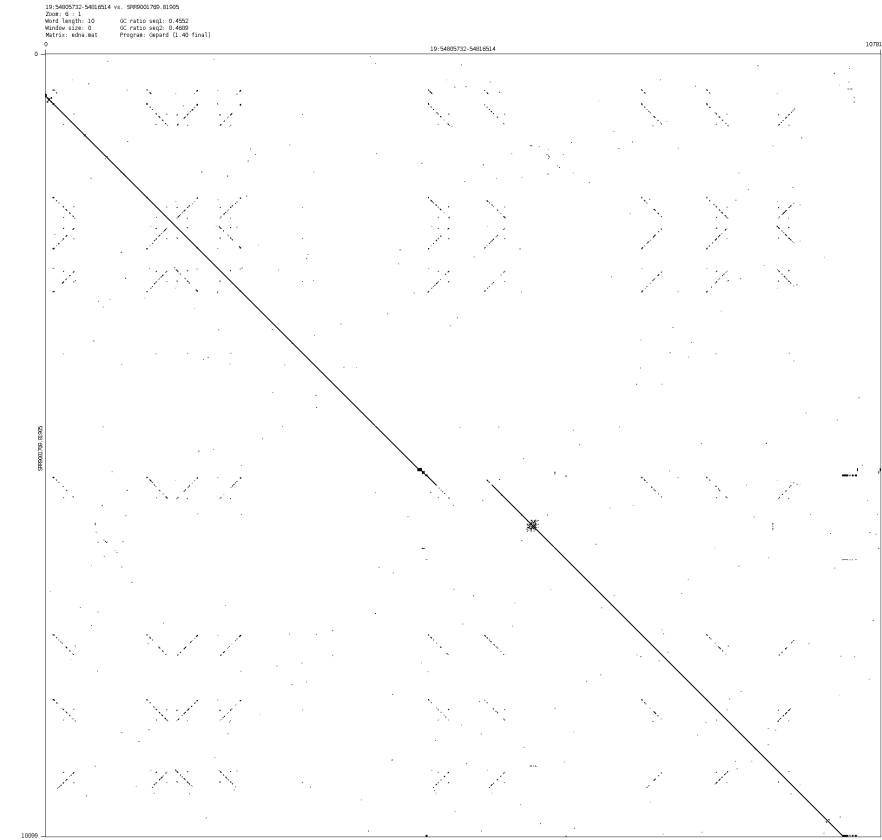




chr18:75,266,983-75,268,167



❖ chr19:54,810,732-54,811,514





chr21:30,552,224-30,552,588

