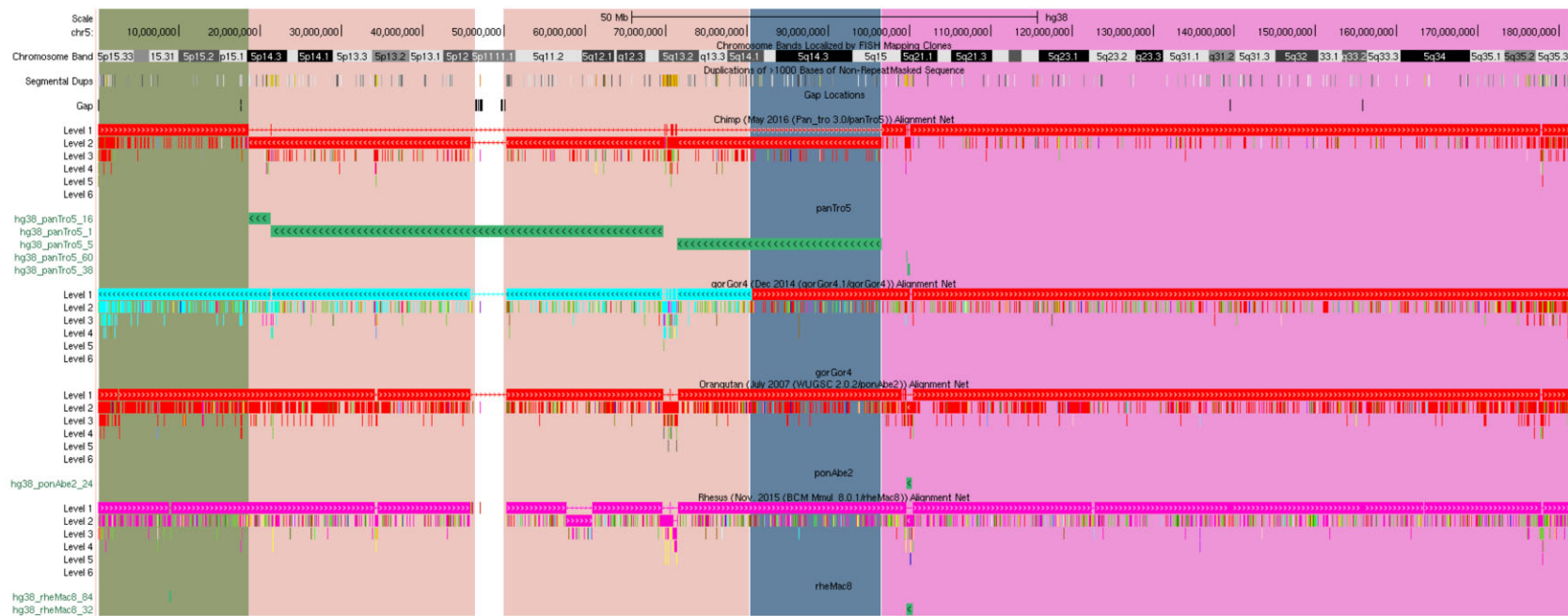
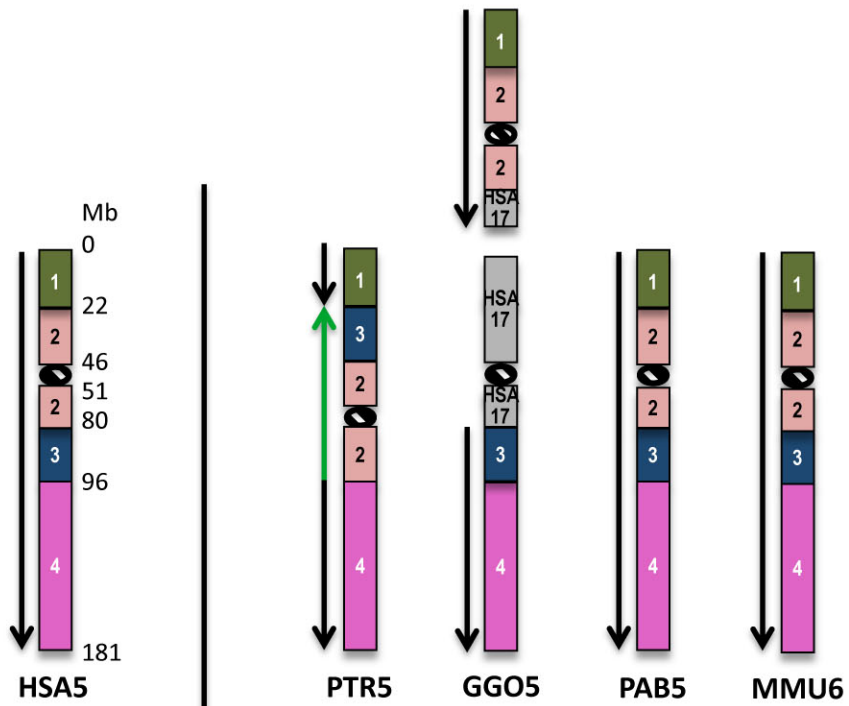
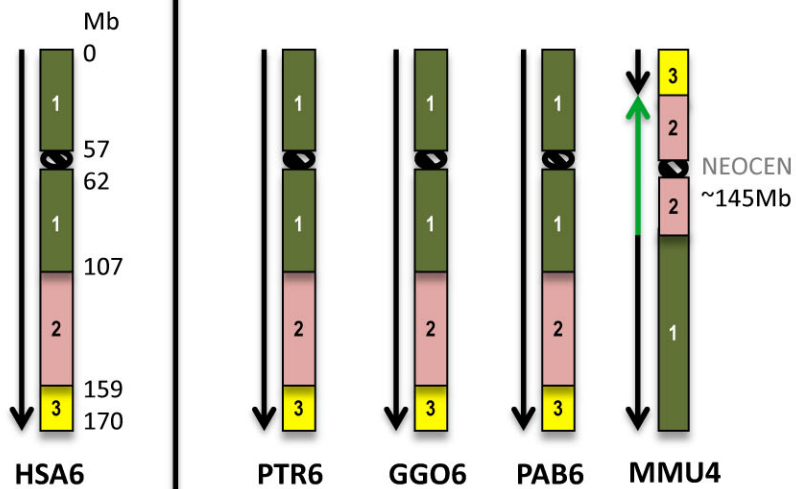
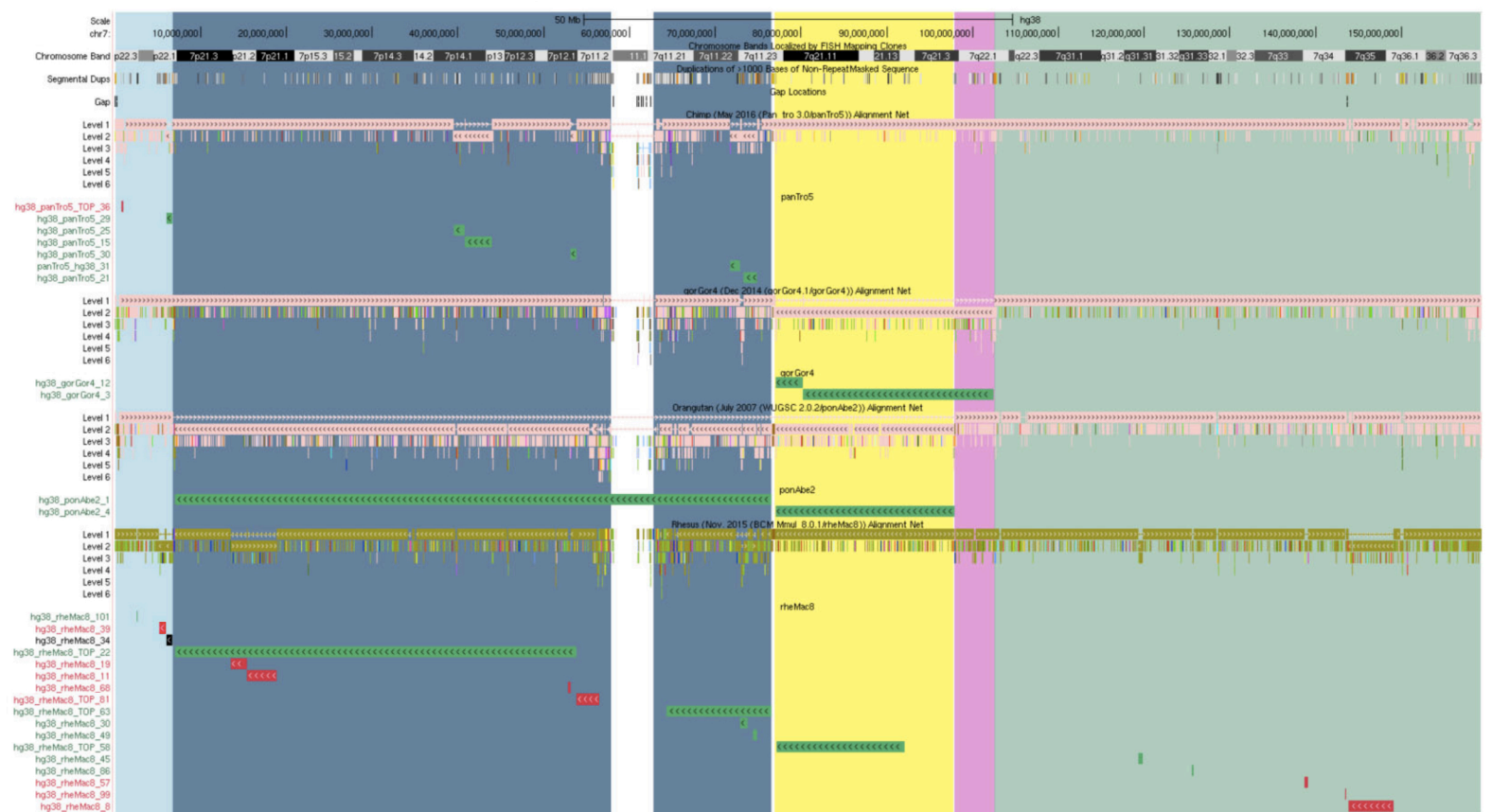
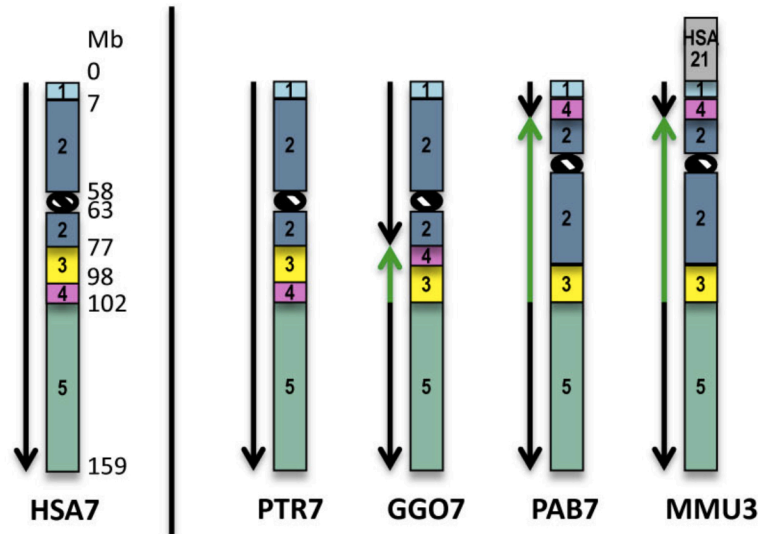
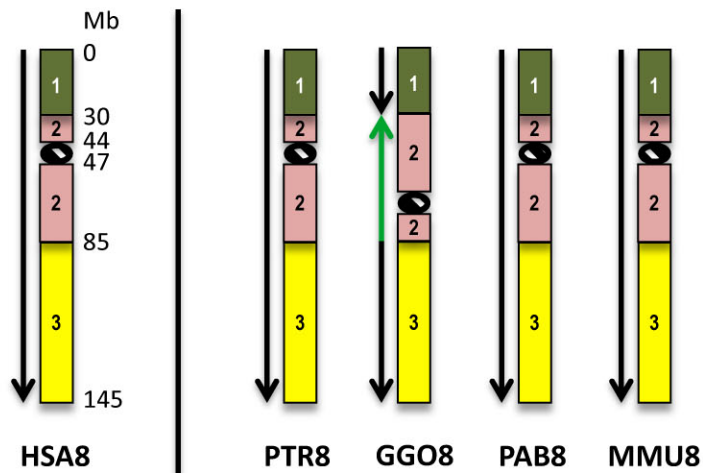


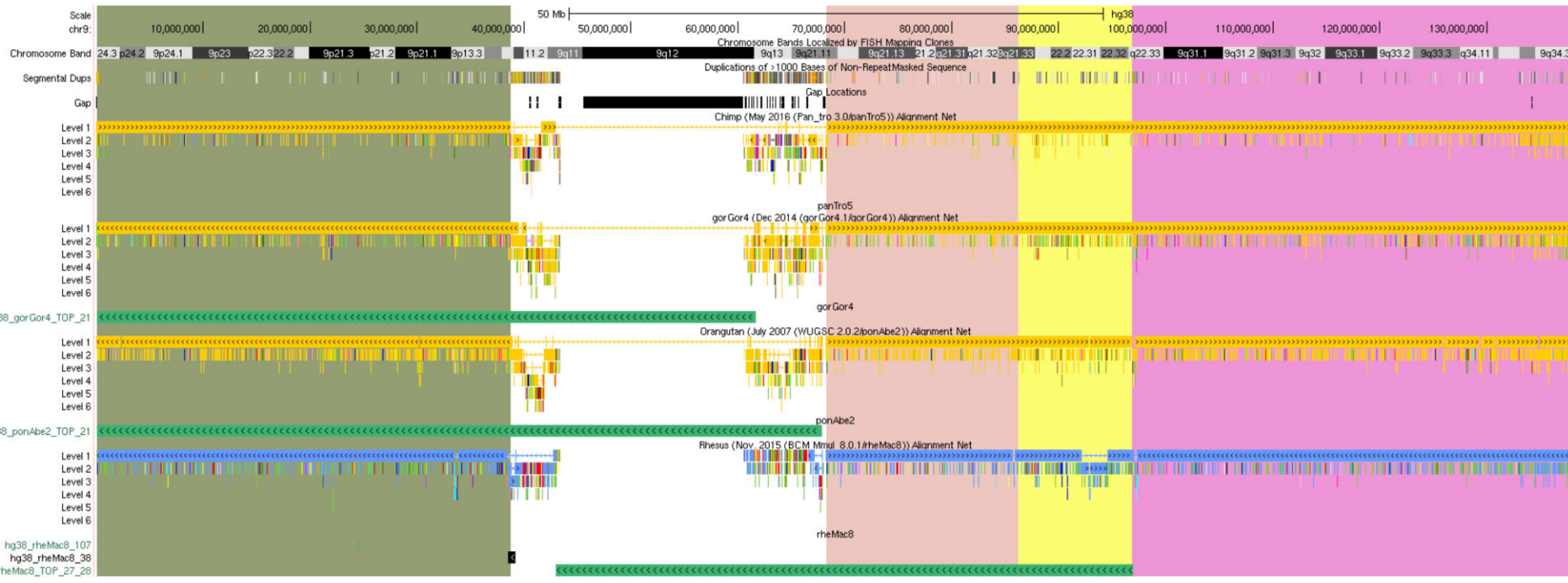
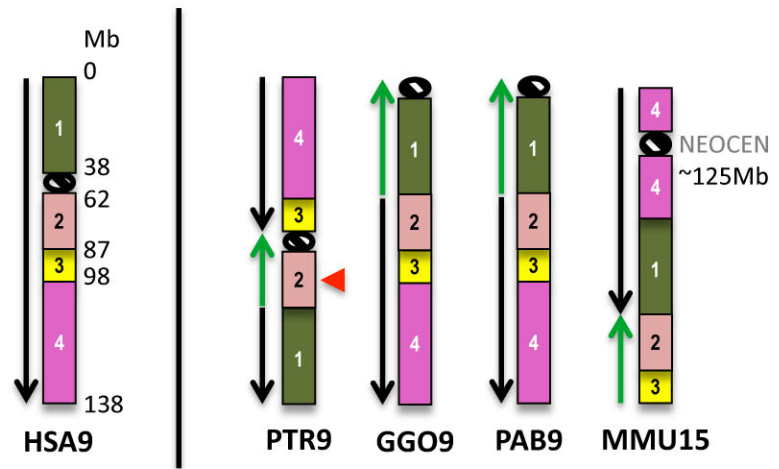
GGO17

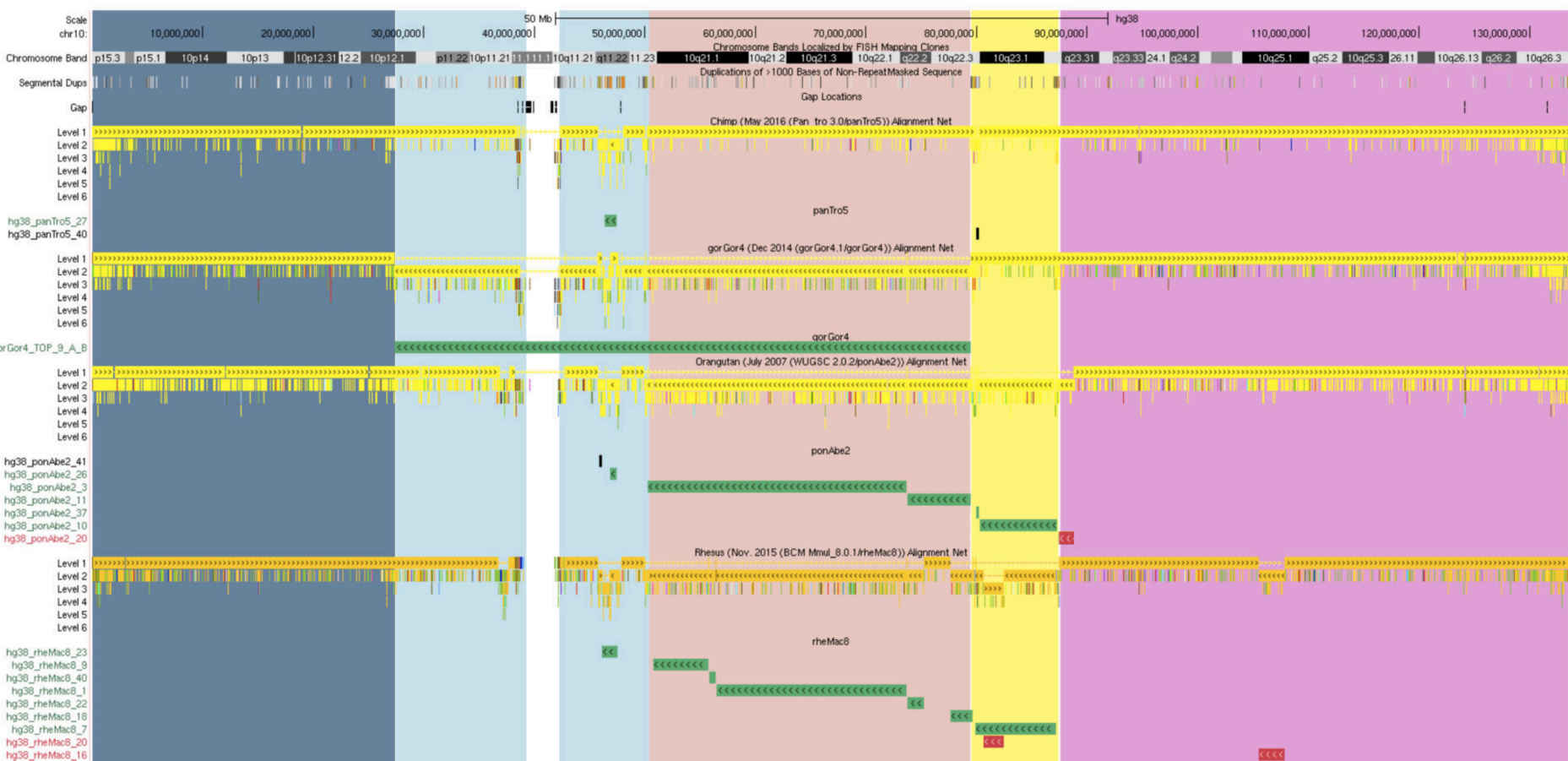
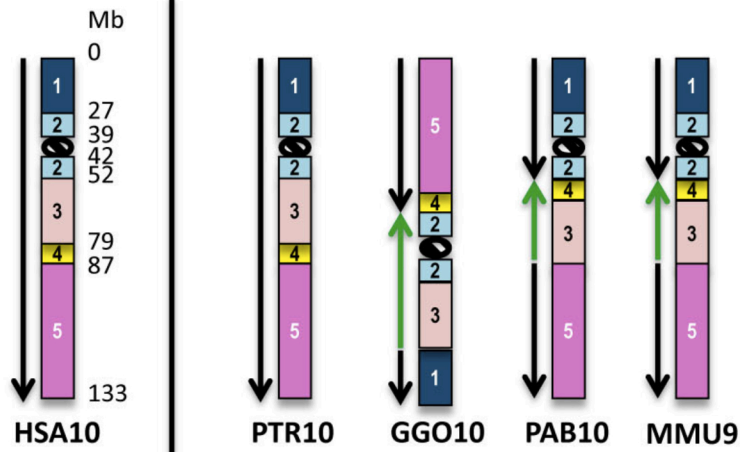


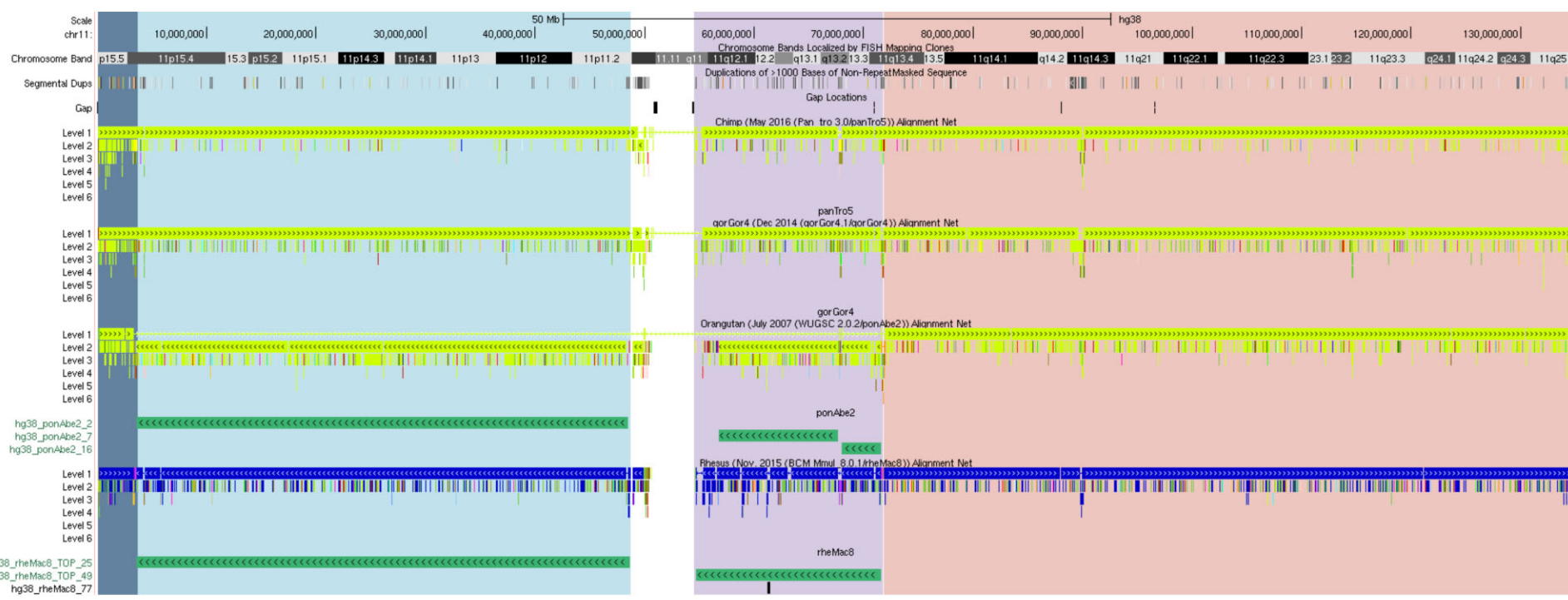
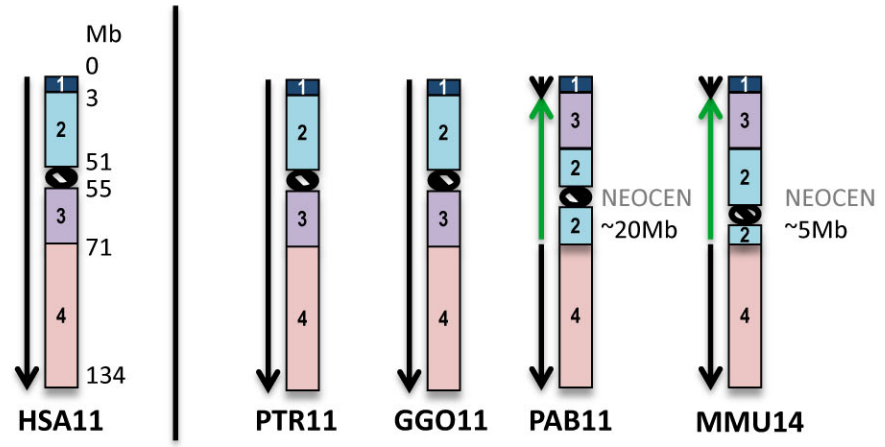


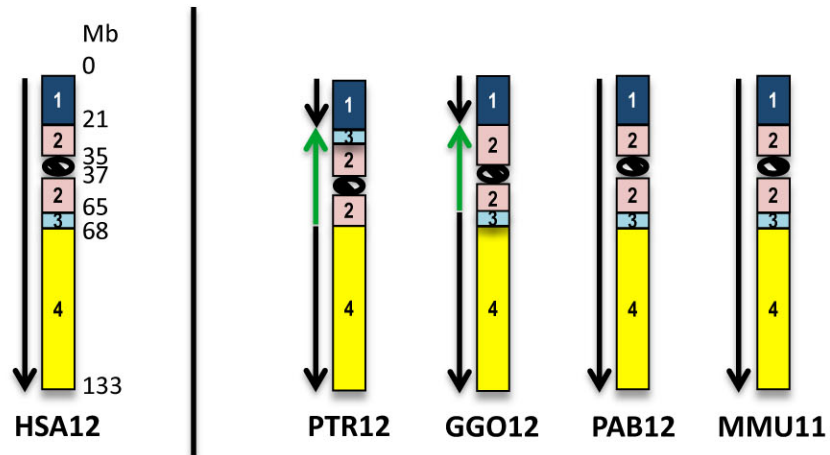


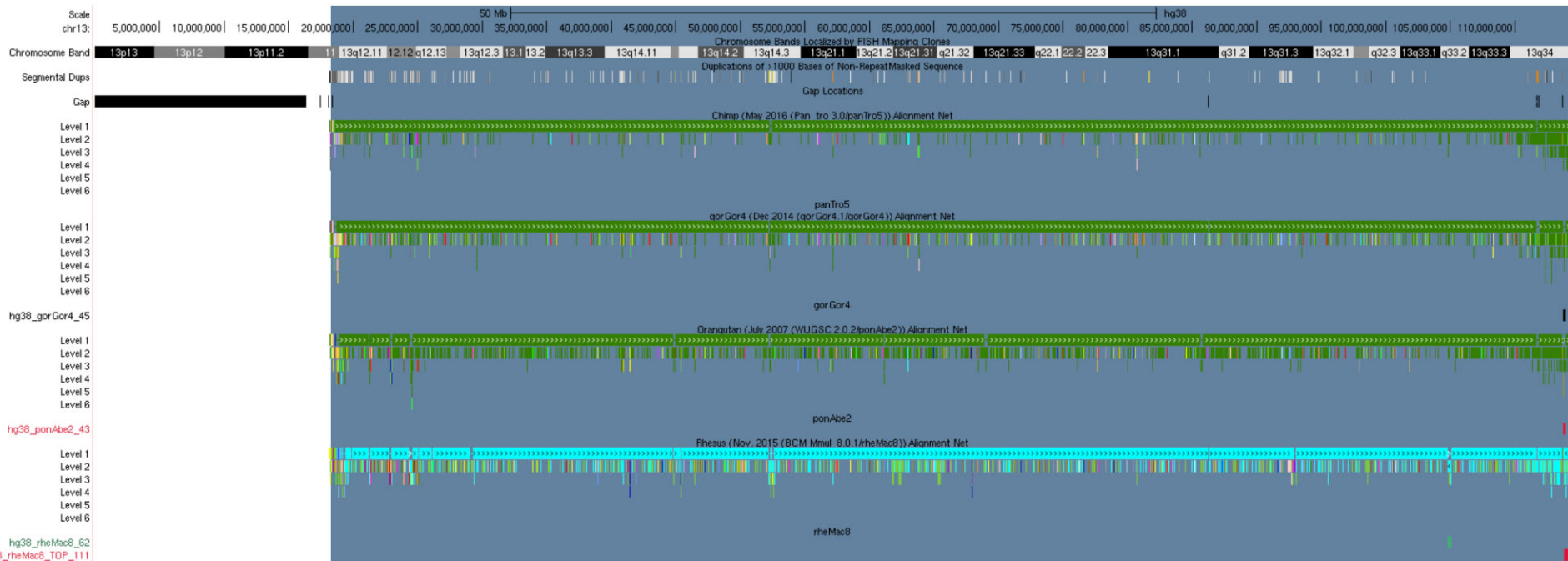
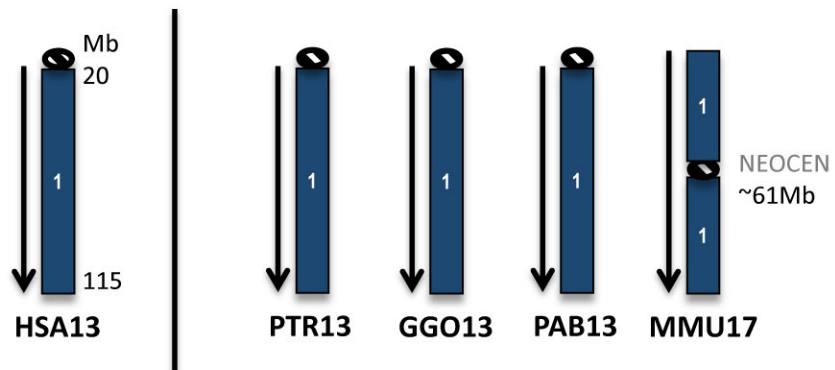


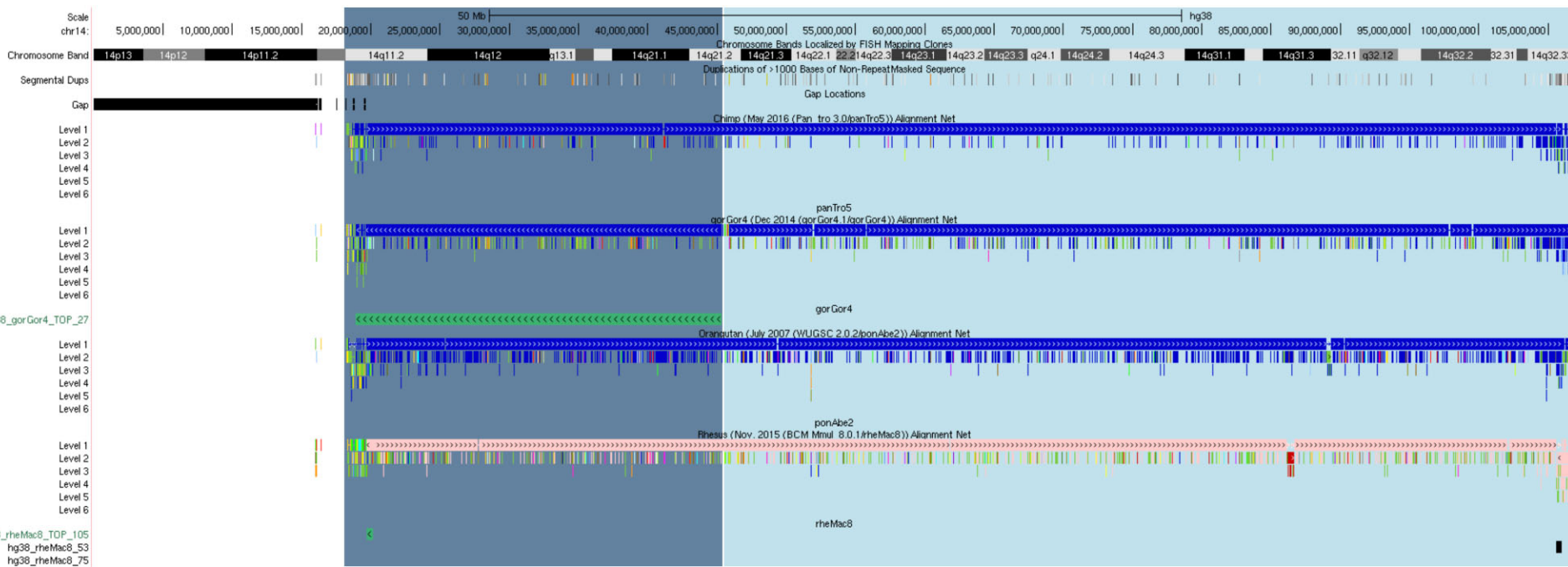
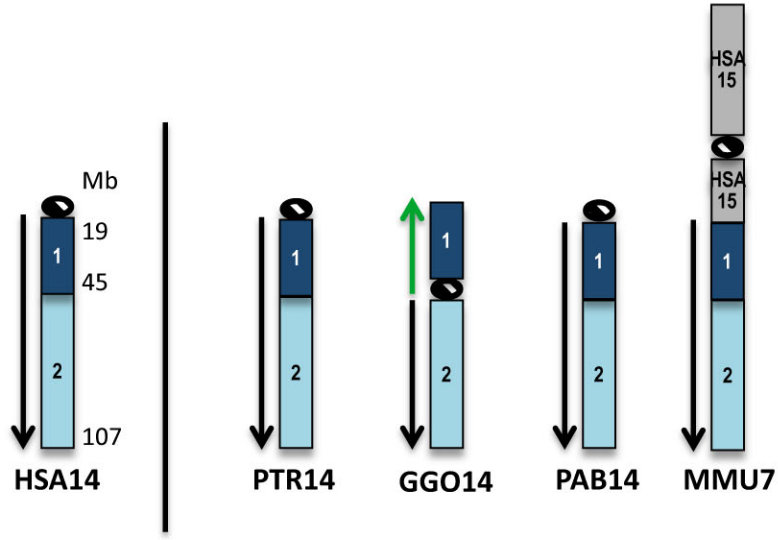


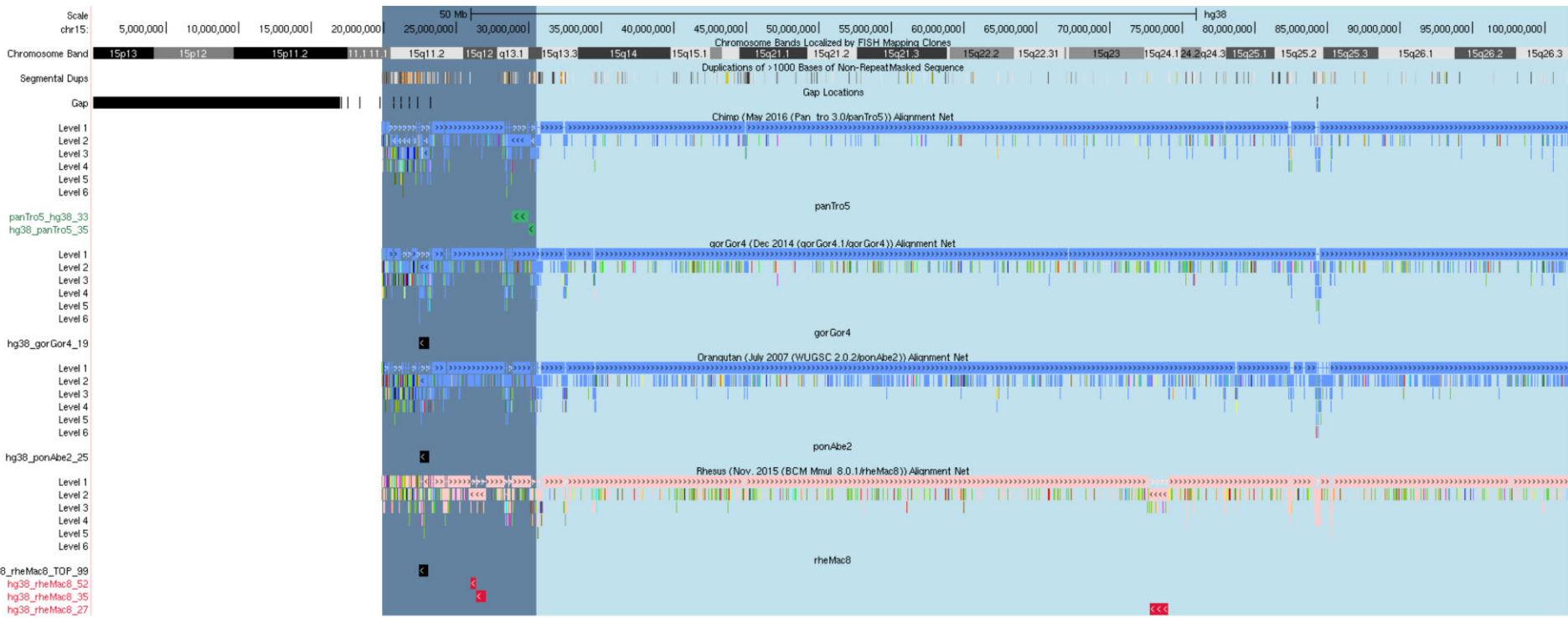
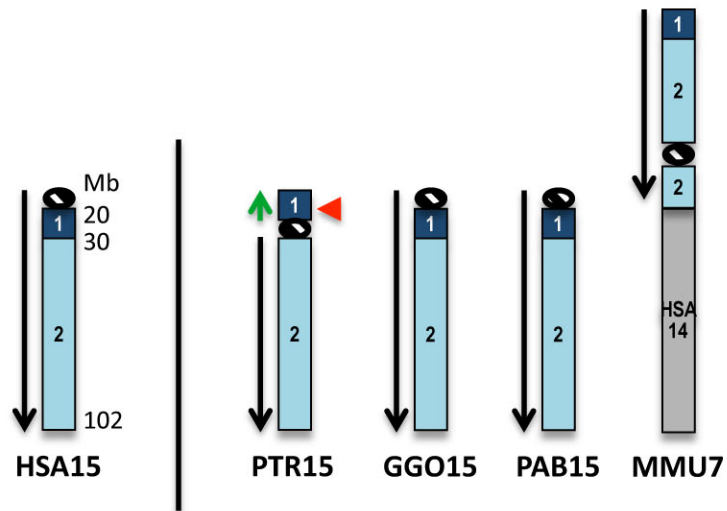


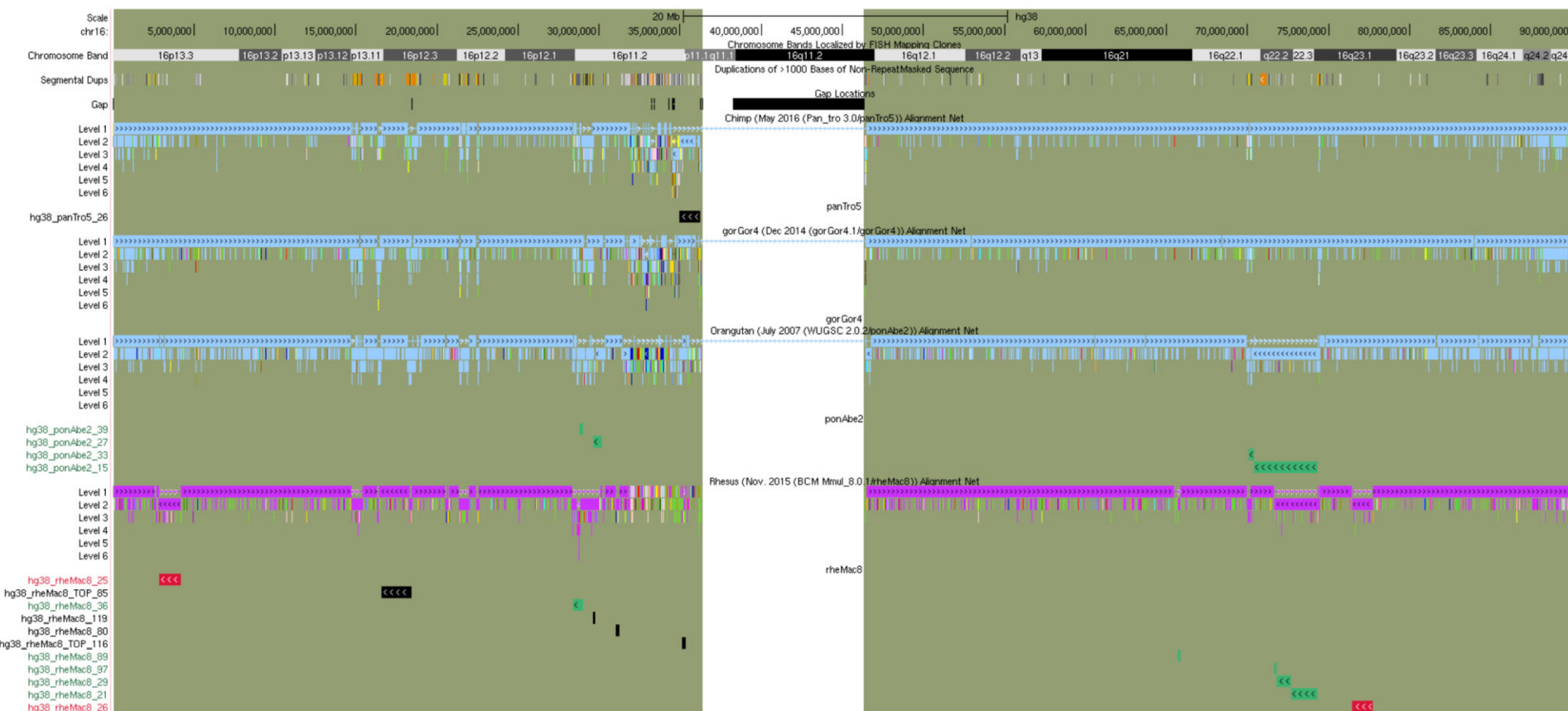
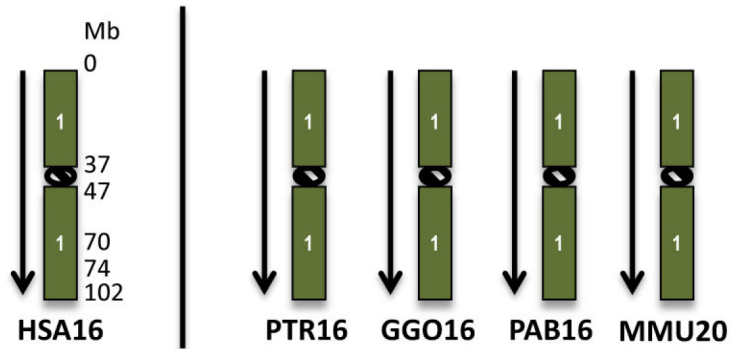




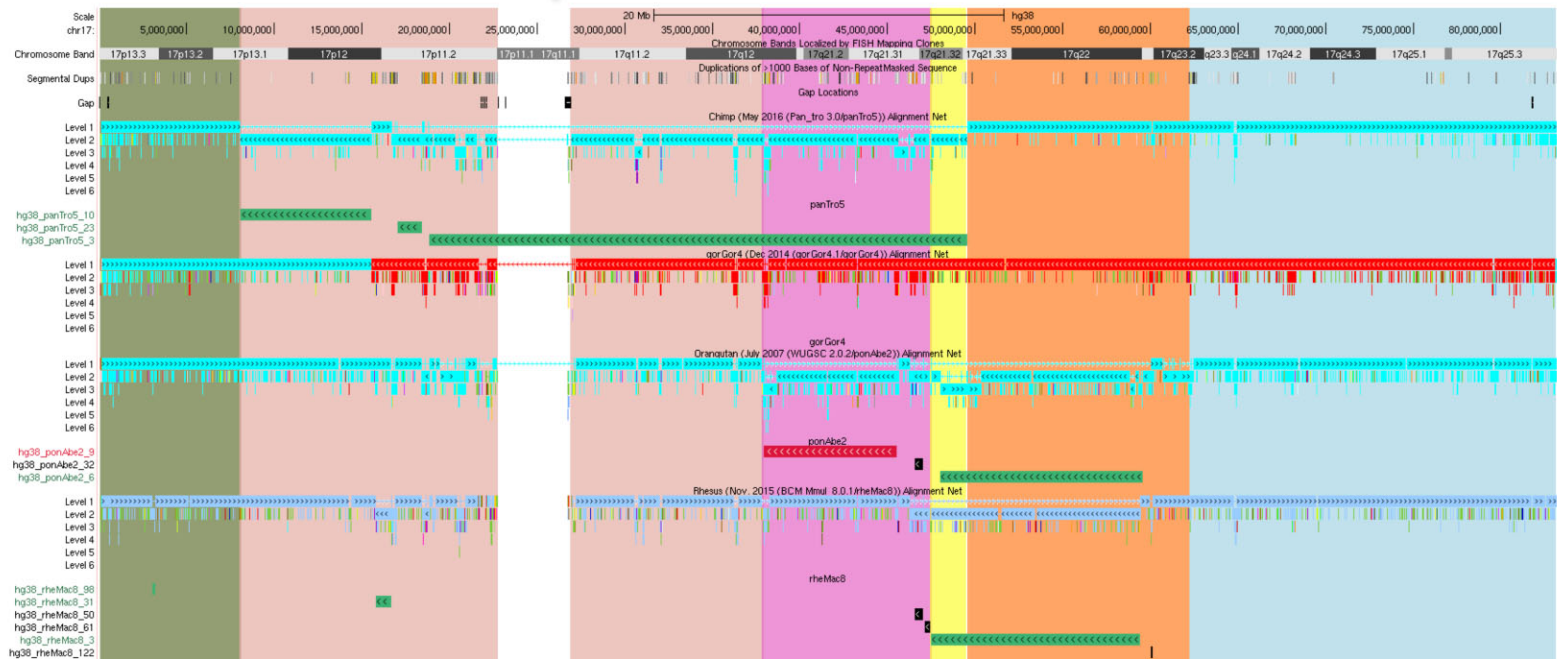
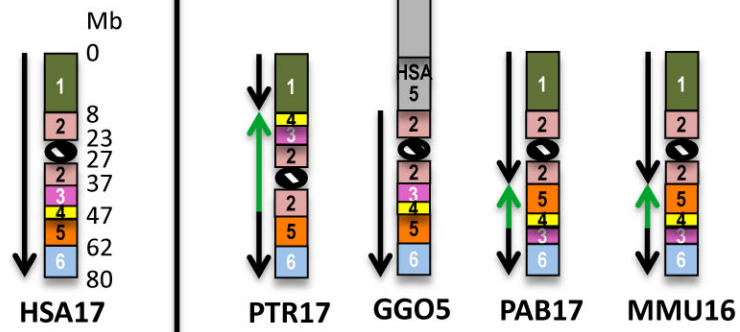


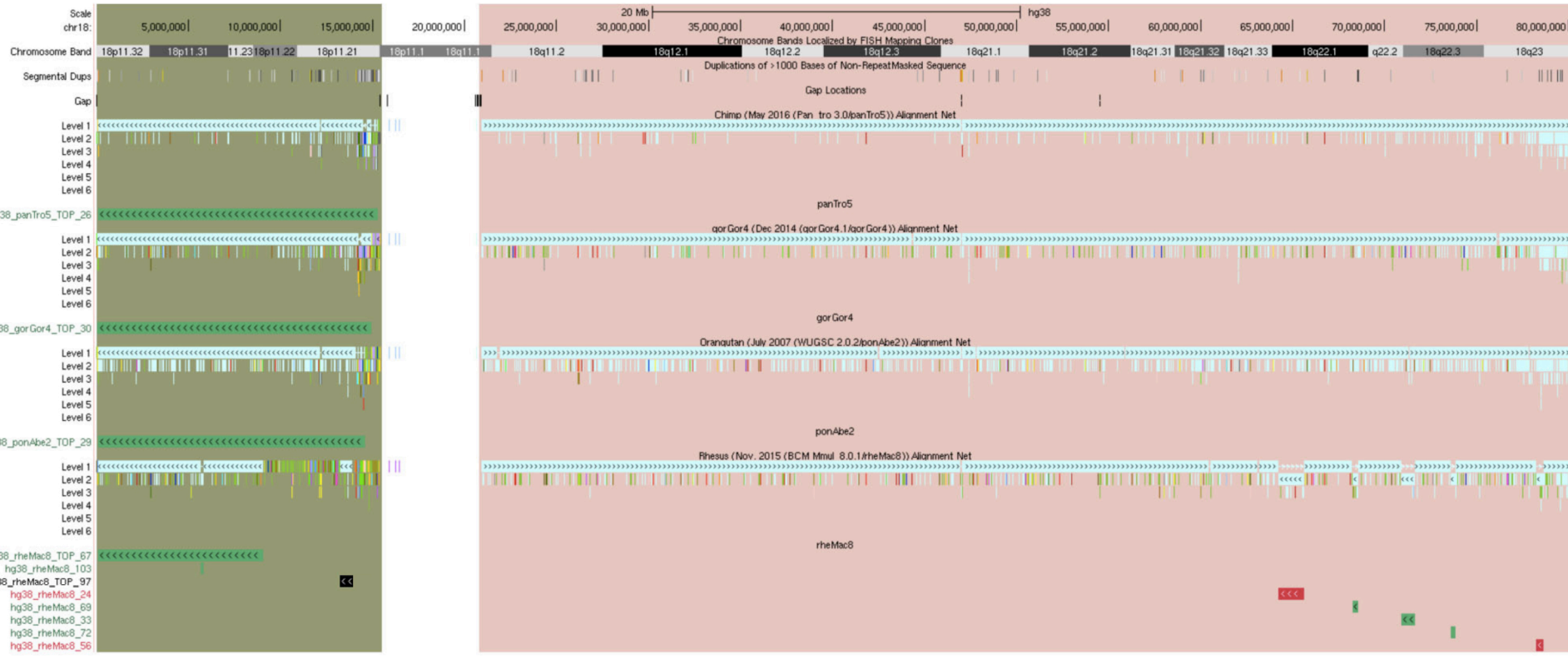
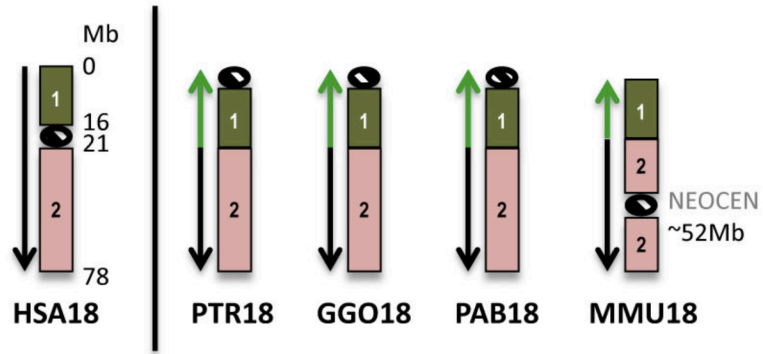


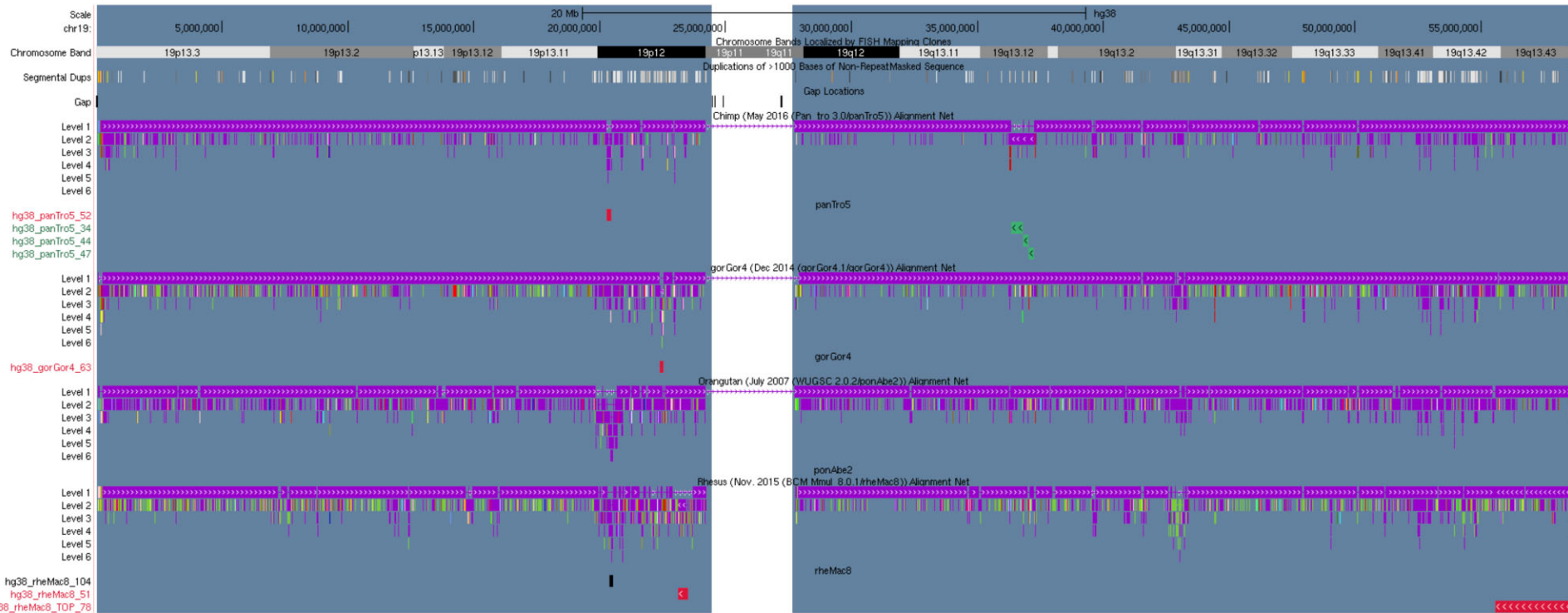
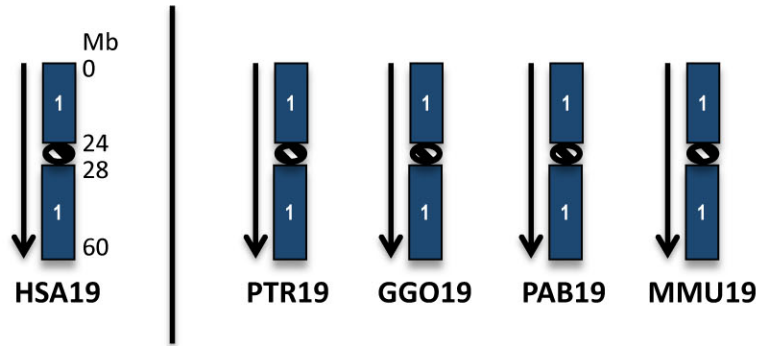


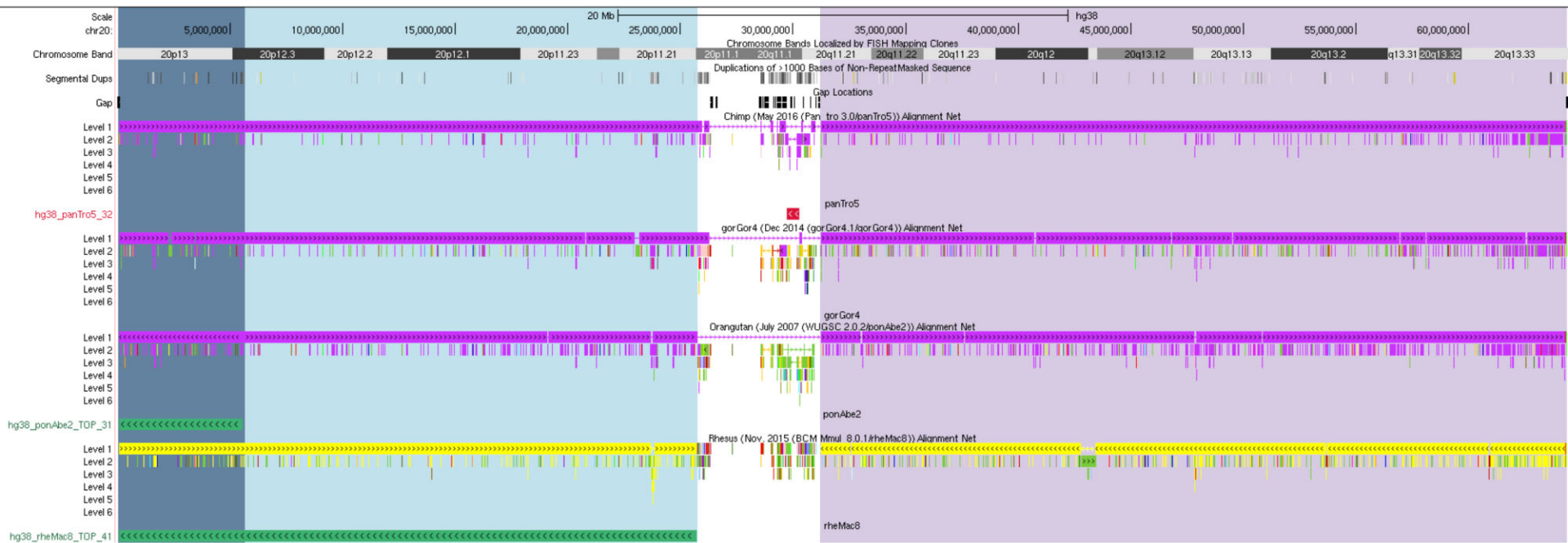
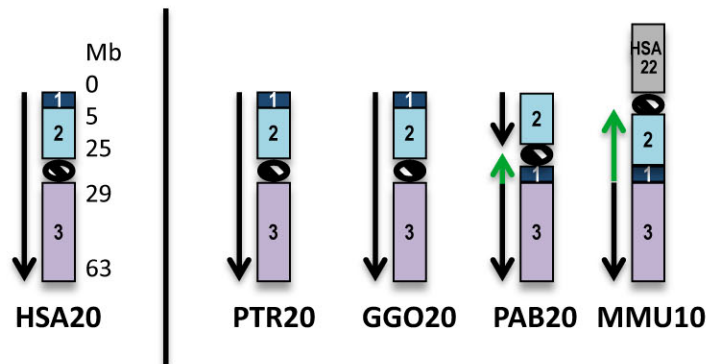


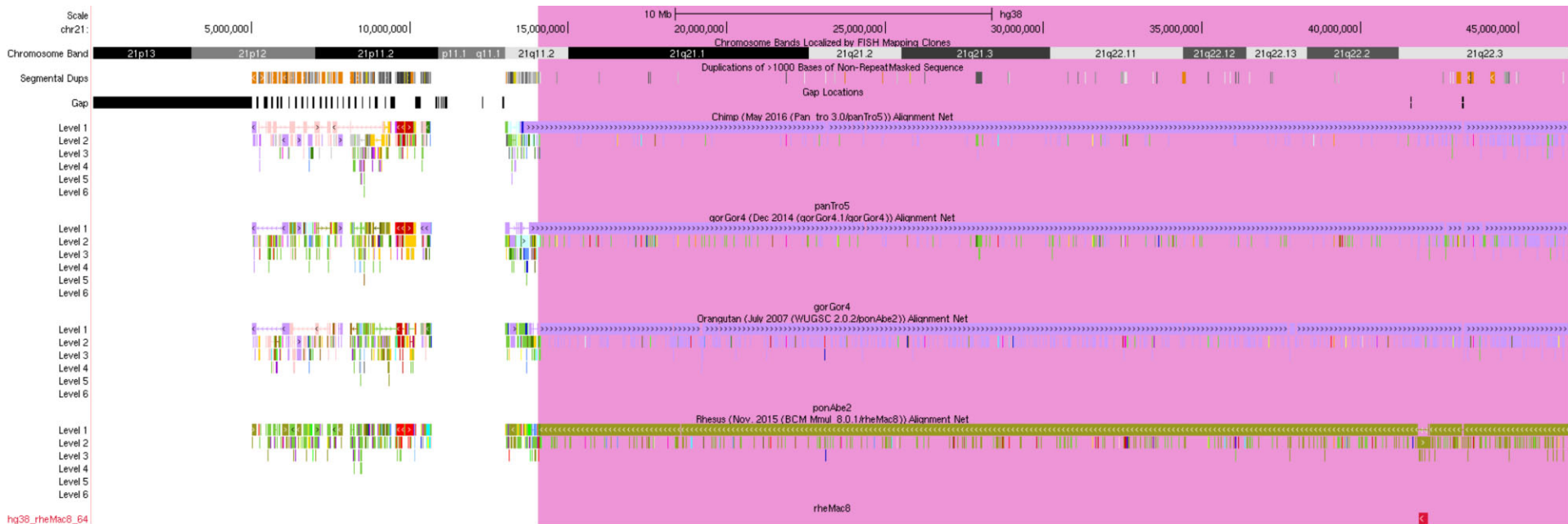
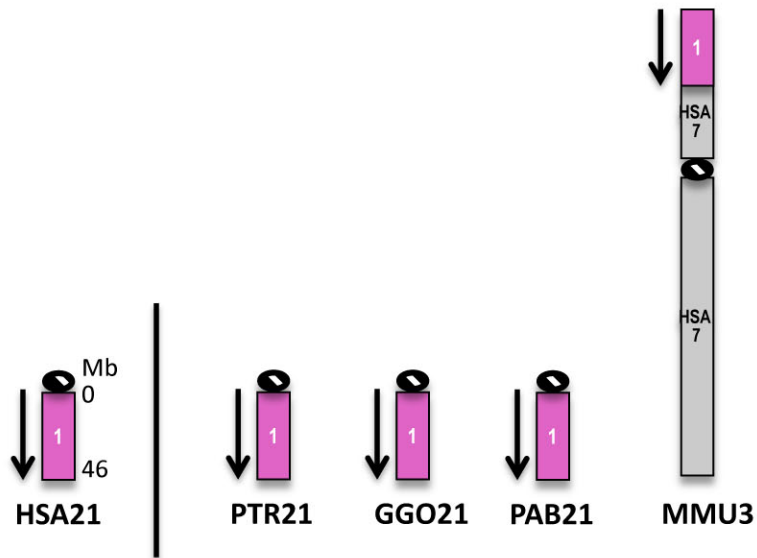
GGO17

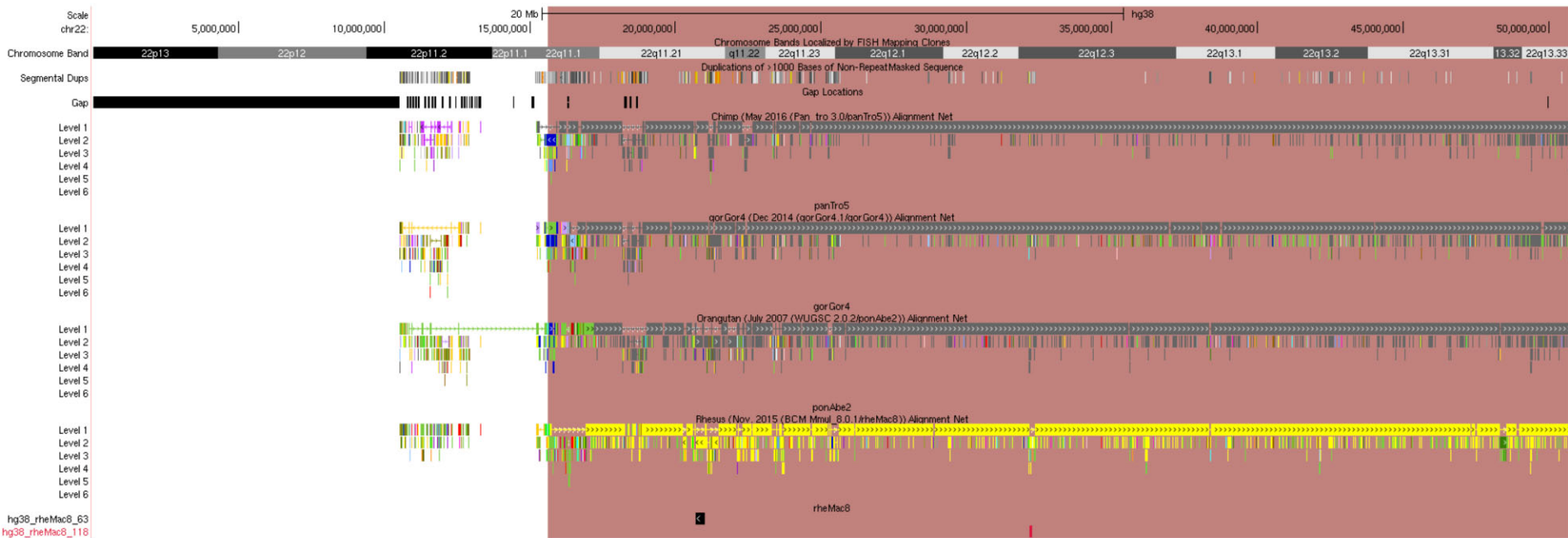
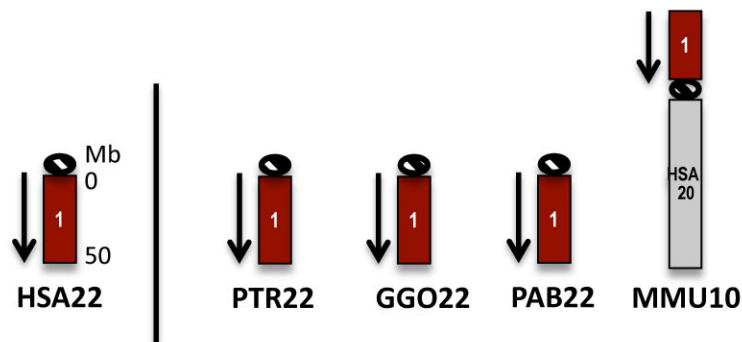












Supplemental Fig. S1. Map of new and previously described genomic inversions.

Chromosome ideograms for human, chimpanzee, gorilla, orangutan and macaque together with the UCSC Genome Browser views of the inversions detected in the study. Ideograms only show previously reported inversions larger than 5 Mb. Synteny blocks, distinguished by colors and numbers, represent regions that are inverted in a least one of the species analyzed, but the order of the markers within the block is conserved in all the different species. Green arrows on the left of each ideogram indicate inverted blocks with respect to human orientation. Small red arrows on the right of the ideograms represent inversions known from previous studies but not detected by net alignments. In the bottom panel, UCSC Genome Browser views of the net alignments and inversions predicted in the present study between human and nonhuman primate genomes. Regions called to be inverted in the present study are shown as green, red and black horizontal bars, and represent real, false and not determined (ND) inversions, respectively. Synteny block colors are consistent with upper panel, and allow to compare regions found to be inverted in the present study with previously identified inversions.

Scale

10 Mb

hg38

Chr1:

110,000,000

115,000,000

120,000,000

125,000,000

130,000,000

135,000,000

140,000,000

145,000,000

Chromosome Bands Localized by FISH Mapping Clones

Chromosome Band

1p21.1

1p13.3

1p13.2

p13.1

1p12

11.2p

11.11q

11q11

1q12

1q21.1

Chr1_inv2

Chr1_inv2



metaphase FISH probes

G248P89047B7



G248P82996H9



Gap Locations

Gap



Duplications of >1000 Bases of Non-RepeatMasked Sequence

Segmental Dups



Scale

1 Mb

hg38

Chr1:

146,000,000

146,500,000

147,000,000

147,500,000

148,000,000

148,500,000

Chromosome Bands Localized by FISH Mapping Clones

Chromosome Band

1q21.1

1q21.2

Chr1_inv3

Chr1_inv3



interphase FISH probes

G248P87580A1



G248P85471H3



G248P86490B5



Gap Locations

Gap

Duplications of >1000 Bases of Non-RepeatMasked Sequence

Segmental Dups





Chromosome Bands Localized by FISH Mapping Clones



Chr10_inv3



interphase FISH probes

WI2-1699F23

WI2-2629B2

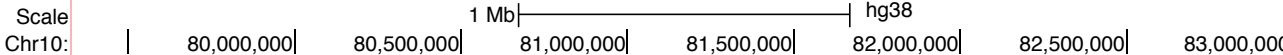
WI2-1673D12

Gap Locations

Gap

Duplications of >1000 Bases of Non-RepeatMasked Sequence

Segmental Dups



Chromosome Bands Localized by FISH Mapping Clones



Chr10_inv4

Chr10_inv4



interphase FISH probes

RP11-1030I16
RP11-319L5
RP11-690O22

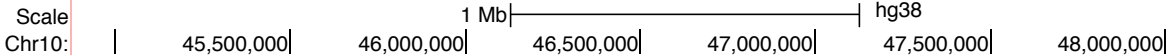


Gap Locations

Duplications of >1000 Bases of Non-RepeatMasked Sequence

Segmental Dups





Chromosome Bands Localized by FISH Mapping Clones



Chr10_inv6

Chr10_inv6



interphase FISH probes

G248P86891H2
 G248P800478D10
 G248P89216B1



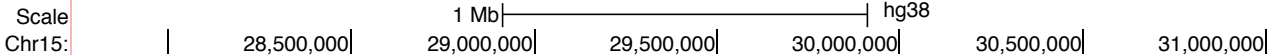
Gap Locations



Duplications of >1000 Bases of Non-RepeatMasked Sequence

Segmental Dups





Gap Locations



Scale 500 kb hg38
Chr15: 73,000,000 73,500,000 74,000,000

Chromosome Bands Localized by FISH Mapping Clones

Chromosome Band

15q24.1



Chr15_inv2

Chr15_inv2



interphase FISH probes

WI2-1551G5



WI2-3701P1



WI2-1763I5

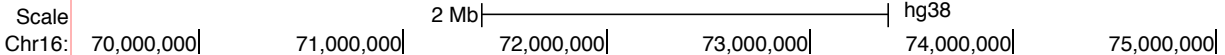


Gap Locations

Duplications of >1000 Bases of Non-RepeatMasked Sequence

Segmental Dups





RP11-779G13 >>

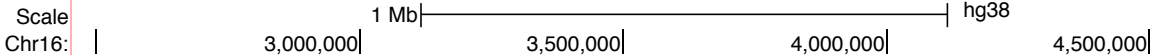
WI2-2368K2

G248P85068H6

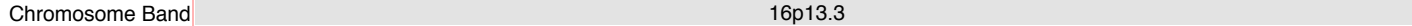


Gap Locations





Chromosome Bands Localized by FISH Mapping Clones



Chr16_inv4

Chr16_inv4



interphase FISH probes

RP11-619M1



RP11-876F24



RP11-707M17



Gap Locations

Gap

Duplications of >1000 Bases of Non-RepeatMasked Sequence

Segmental Dups

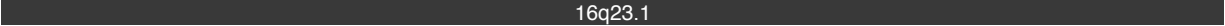


Scale 500 kb hg38
Chr16: 76,500,000 77,000,000 77,500,000

Chromosome Bands Localized by FISH Mapping Clones

Chromosome Band

16q23.1



Chr16_inv6

Chr16_inv6



interphase FISH probes

WI2-1991O15



WI2-1773I19



WI2-3622O10



Gap Locations

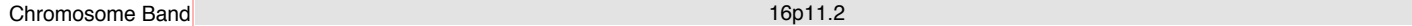
Gap

Duplications of >1000 Bases of Non-RepeatMasked Sequence

Segmental Dups



Chromosome Bands Localized by FISH Mapping Clones



Chr16_inv8



G248P82830F11
 G248P81405C1
 G248P80035A4

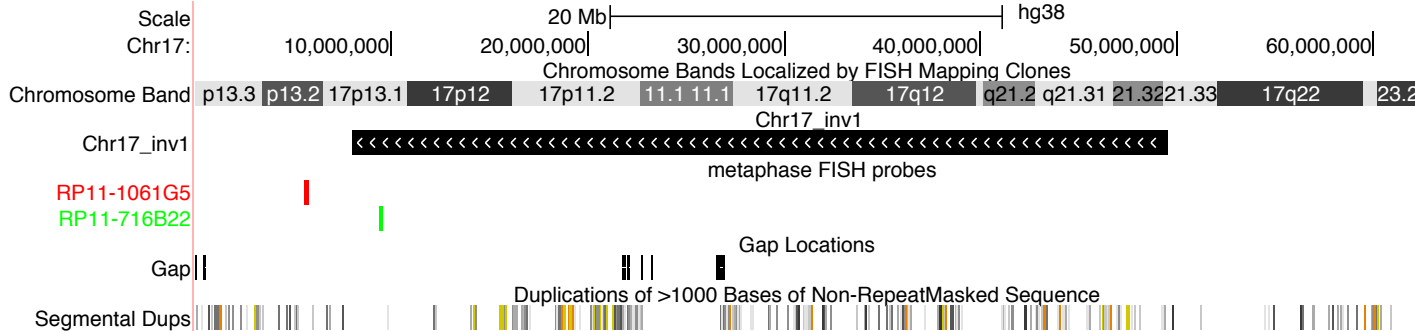


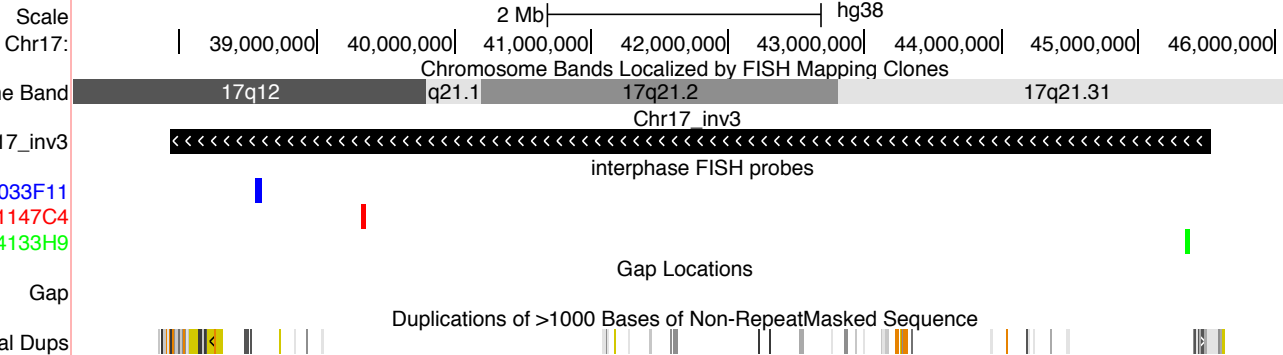
Gap Locations

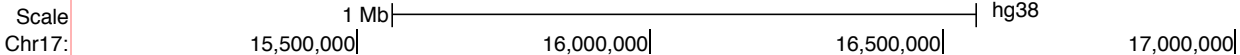
Duplications of >1000 Bases of Non-RepeatMasked Sequence

Segmental Dups









Chromosome Bands Localized by FISH Mapping Clones



Chr17_inv4



Gap Locations



Duplications of >1000 Bases of Non-RepeatMasked Sequence





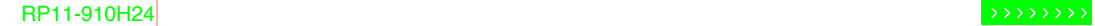
Chromosome Bands Localized by FISH Mapping Clones



Chr18_inv2



interphase FISH probes



Gap Locations

Duplications of >1000 Bases of Non-RepeatMasked Sequence





Chromosome Bands Localized by FISH Mapping Clones



Chr19_inv2

Chr19_inv2



interphase FISH probes

RP11-1148D20



RP11-1088H16



RP11-587I9



Gap Locations

Gap

Duplications of >1000 Bases of Non-RepeatMasked Sequence

Segmental Dups



Scale

2 Mb

hg38

Chr4:

4,000,000

5,000,000

6,000,000

7,000,000

8,000,000

9,000,000

10,000,000

Chromosome Bands Localized by FISH Mapping Clones

Chromosome Band

4p16.3

4p16.2

4p16.1

Chr4_inv5

Chr4_inv5



metaphase FISH probes

G248P8419H5



G248P89041F10



Gap Locations

Gap

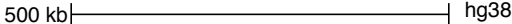


Duplications of >1000 Bases of Non-RepeatMasked Sequence

Segmental Dups



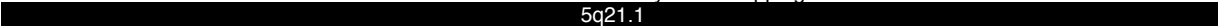
Scale



Chr5: 99,500,000 |

Chromosome Bands Localized by FISH Mapping Clones

Chromosome Band



Chr5_inv2



interphase FISH probes

RP11-350L5



RP11-467C9



RP11-368A20



Gap Locations

Gap

Duplications of >1000 Bases of Non-RepeatMasked Sequence

Segmental Dups



Scale

1 Mb hg38

Chr6:

104,000,000 | 104,500,000 | 105,000,000 | 105,500,000 | 106,000,000 | 106,500,000 | 107,000,000 | 107,500,000

Chromosome Bands Localized by FISH Mapping Clones

Chromosome Band

6q16.3

6q21

Chr6_inv2

Chr6_inv2



interphase FISH probes

WI2-2840C22



WI2-1971C10



WI2-2534L10



Gap Locations

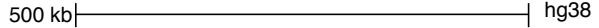
Gap

Duplications of >1000 Bases of Non-RepeatMasked Sequence

Segmental Dups



Scale



Chr7:

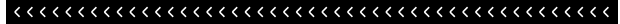
Chromosome Bands Localized by FISH Mapping Clones

Chromosome Band



Chr7_inv10

Chr7_inv10



interphase FISH probes

RP11-1056A8



RP11-775N3



RP11-478G18



Gap Locations

Gap

Duplications of >1000 Bases of Non-RepeatMasked Sequence

Segmental Dups



Scale

2 Mb

hg38

Chr7:

14,000,000

15,000,000

16,000,000

17,000,000

18,000,000

19,000,000

Chromosome Bands Localized by FISH Mapping Clones

Chromosome Band

7p21.3

7p21.2

7p21.1

Chr7_inv3

Chr7_inv3



interphase FISH probes

WI2-455G21



WI2-3496M23



WI2-2240K12



Gap Locations

Gap

Duplications of >1000 Bases of Non-RepeatMasked Sequence

Segmental Dups



Scale

2 Mb

hg38

Chr7:

144,000,000

145,000,000

146,000,000

147,000,000

148,000,000

149,000,000

Chromosome Bands Localized by FISH Mapping Clones

Chromosome Band

7q34

7q35

7q36.1

Chr7_inv4

Chr7_inv4



interphase FISH probes

WI2-491B21



WI2-1692N22



WI2-827H23



Gap Locations

Gap



Duplications of >1000 Bases of Non-RepeatMasked Sequence

Segmental Dups



Scale

1 Mb

hg38

Chr7:

72,000,000

72,500,000

73,000,000

73,500,000

74,000,000

74,500,000

75,000,000

75,500,000

Chromosome Bands Localized by FISH Mapping Clones

Chromosome Band

7q11.22

7q11.23

Chr7_inv6

Chr7_inv6



interphase FISH probes

WI2-3210F8



RP11-460F3



RP11-351B3



Gap Locations

Gap

Duplications of >1000 Bases of Non-RepeatMasked Sequence

Segmental Dups



Scale

1 Mb

hg38

Chr8:

50,500,000

51,000,000

51,500,000

52,000,000

52,500,000

53,000,000

53,500,000

Chromosome Bands Localized by FISH Mapping Clones

Chromosome Band

8q11.21

8q11.22

8q11.23

Chr8_inv3

Chr8_inv3



interphase FISH probes

WI2-782F1



WI2-2075A5



WI2-2945D23



Gap Locations

Gap

Duplications of >1000 Bases of Non-RepeatMasked Sequence

Segmental Dups



Scale

1 Mb

hg38

Chr8:

40,500,000

41,000,000

41,500,000

42,000,000

42,500,000

43,000,000

43,500,000

Chromosome Bands Localized by FISH Mapping Clones

Chromosome Band

8p11.21

8p11.1

Chr8_inv4

Chr8_inv4



interphase FISH probes

RP11-1066P10



RP11-939C8



RP11-875E16



Gap Locations

Gap

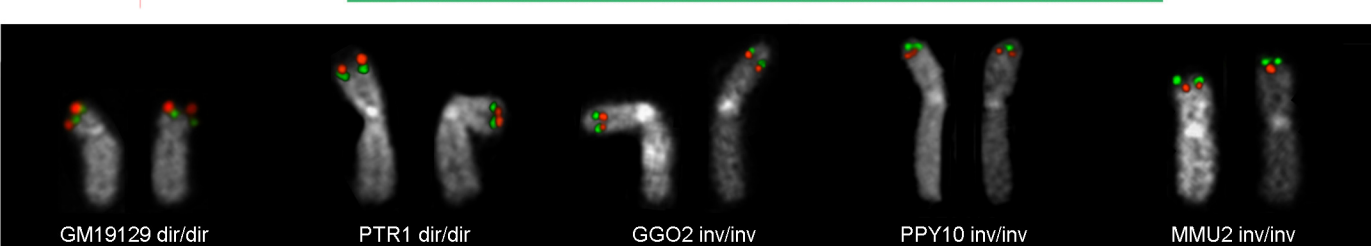
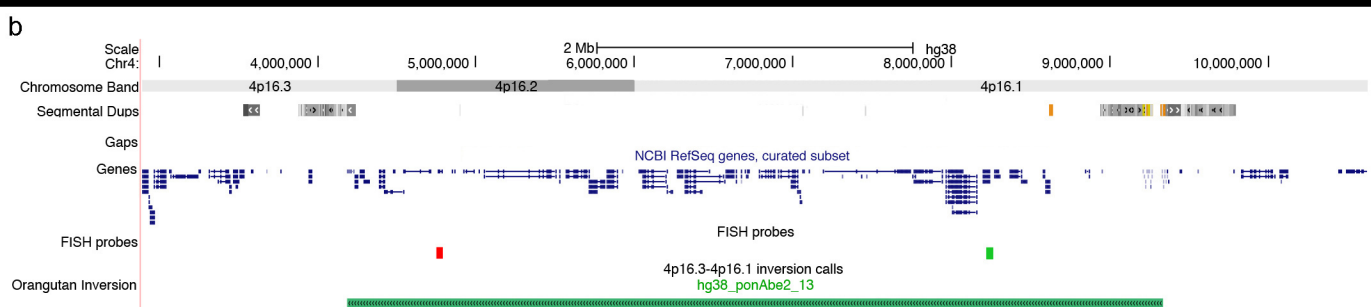
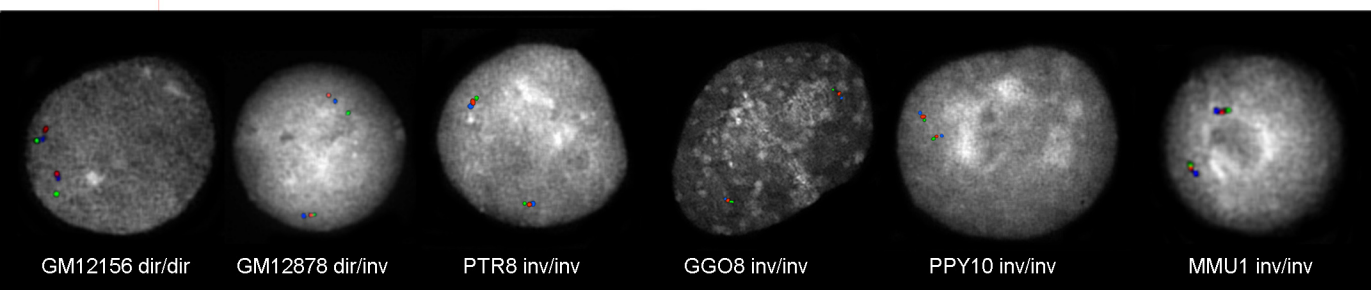
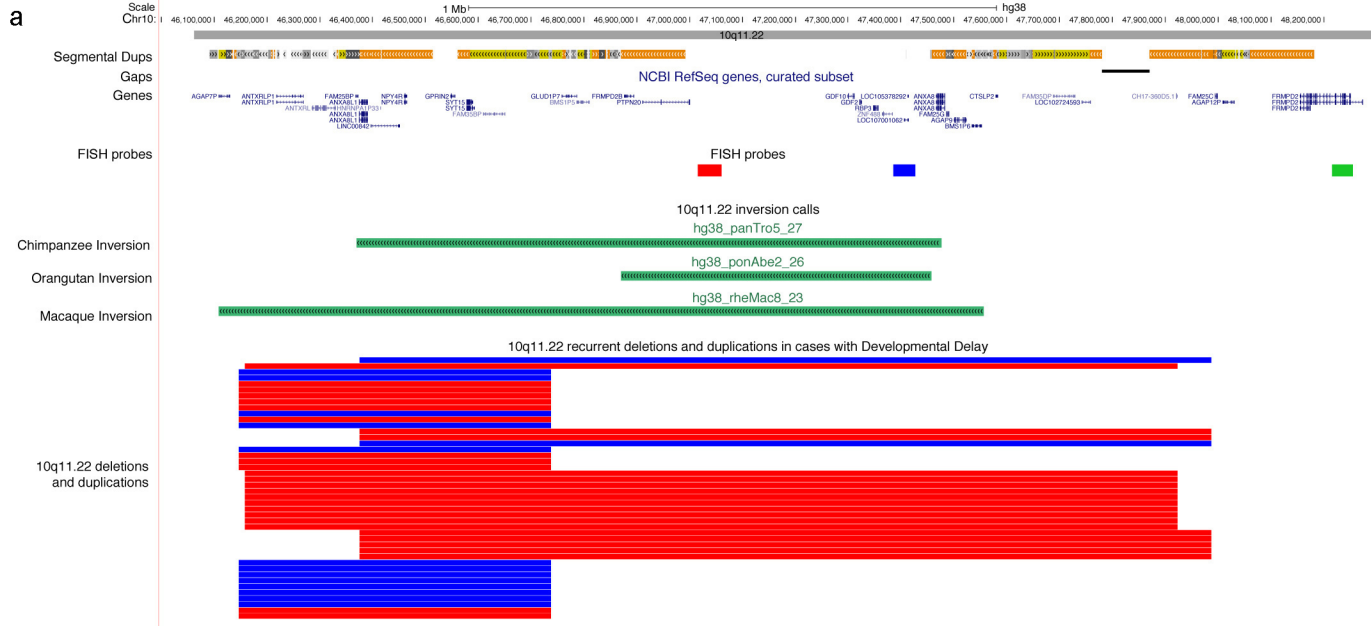
Duplications of >1000 Bases of Non-RepeatMasked Sequence

Segmental Dups



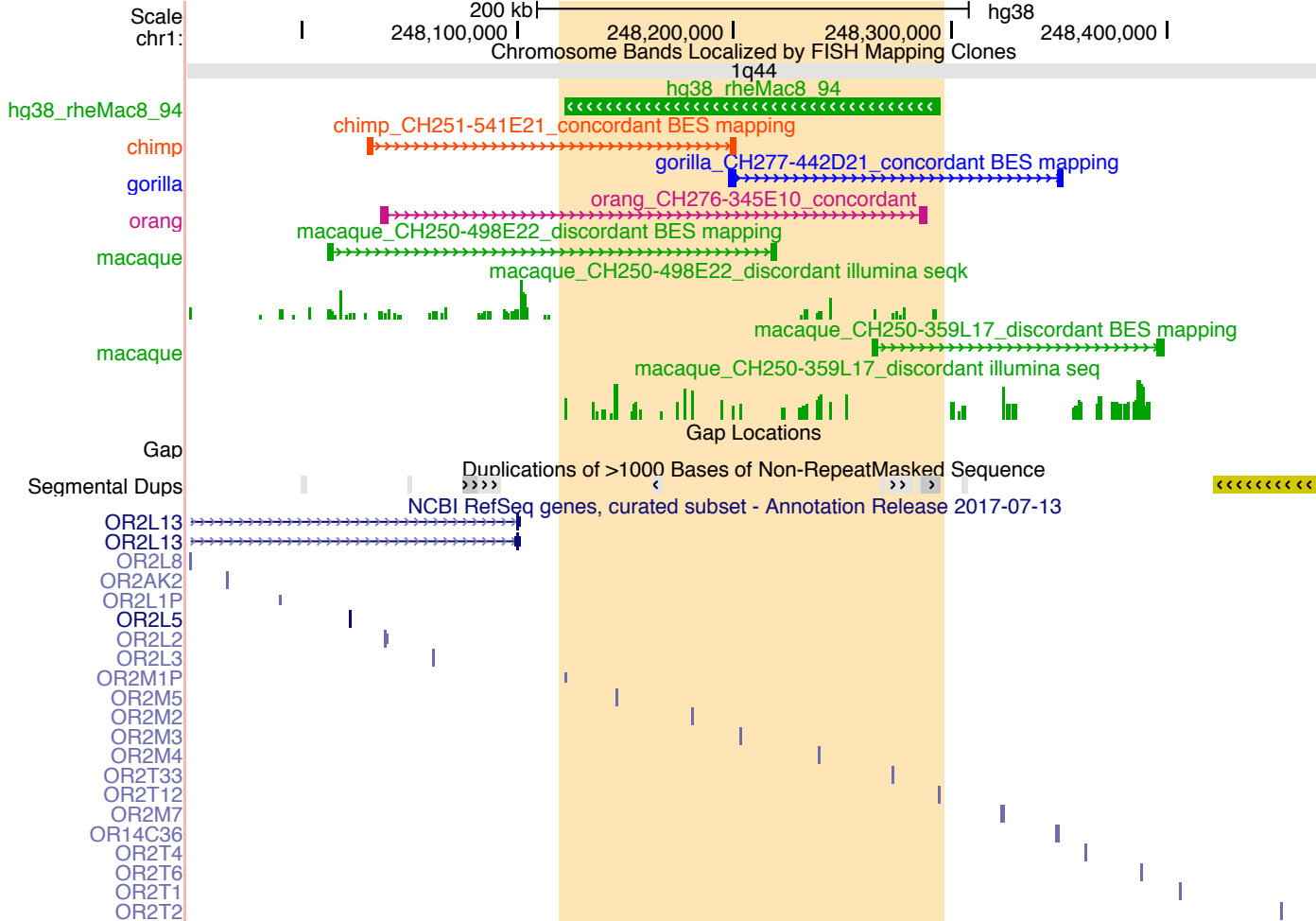
Supplemental Fig. S2. FISH experiments and probe locations.

The location of the probes and the type of FISH experiment (in metaphase or interphase) is shown with respect to the genomic regions studied.



Supplemental Fig. S3. 10q11.22 and 4p16.3-4p16.1 inversion regions.

a) UCSC Genome Browser view of the 10q11.22 region illustrating inversion calls between human and chimpanzee, human and orangutan, and human and macaque. FISH probes used to validate the inversion are shown. Recurrent microdeletions/microduplications are described at the 10q11.22 locus. At the bottom, FISH results on interphase nuclei show that the inversion is polymorphic in human while all the other nonhuman primates tested carry the inverted haplotype. b) UCSC Genome Browser view of the 4p16.3-4p16.1 region illustrating the inversion call between human and macaque. FISH probes used to validate the inversion are shown. At the bottom, FISH results on metaphase nuclei show that the region was is in inverted orientation in gorilla, orangutan and macaque.



Scale chr1: | 194,100,000 | 200 kb | 194,200,000 | 194,300,000 | 194,400,000 | 194,500,000 | hg38

Chromosome Bands | localized by FISH Mapping Clones

1q31.3

hg38_ponAbe2_55

hg38_ponAbe2_55

chimp

chimp_CH251-208F22_concordant BES mapping

chimp_CH251-208F22_concordant illumina seq

gorilla

gorilla_CH277-33N7_concordant BES mapping

gorilla_CH277-33N7_concordant illumina seq

orang

orang_CH276-252K6_discordant BES mapping

orang_CH276-252K6_discordant illumina seq

orang

orang_CH276-176F16_discordant BES mapping

orang_CH276-176F16_discordant illumina seq

macaque

macaque_CH250-343I17_discordant BES mapping

macaque_CH250-343I17_discordant illumina seq

marmoset

marmoset_CH259-53C6_discordant BES mapping

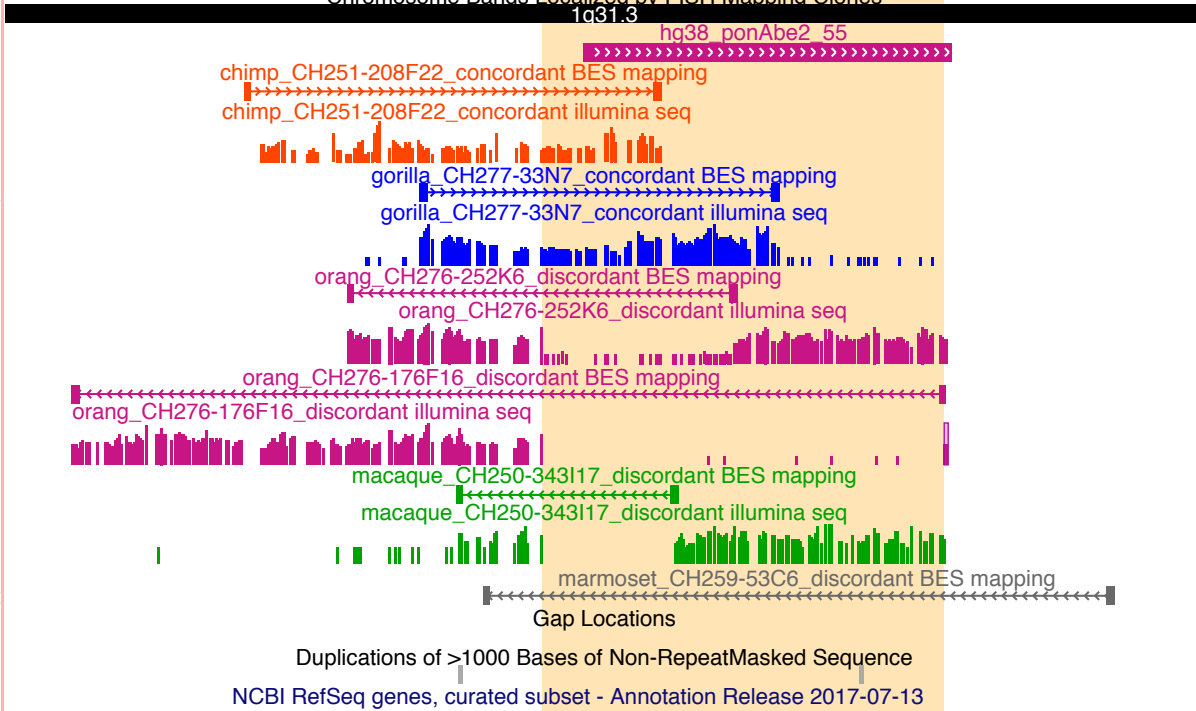
Gap Locations

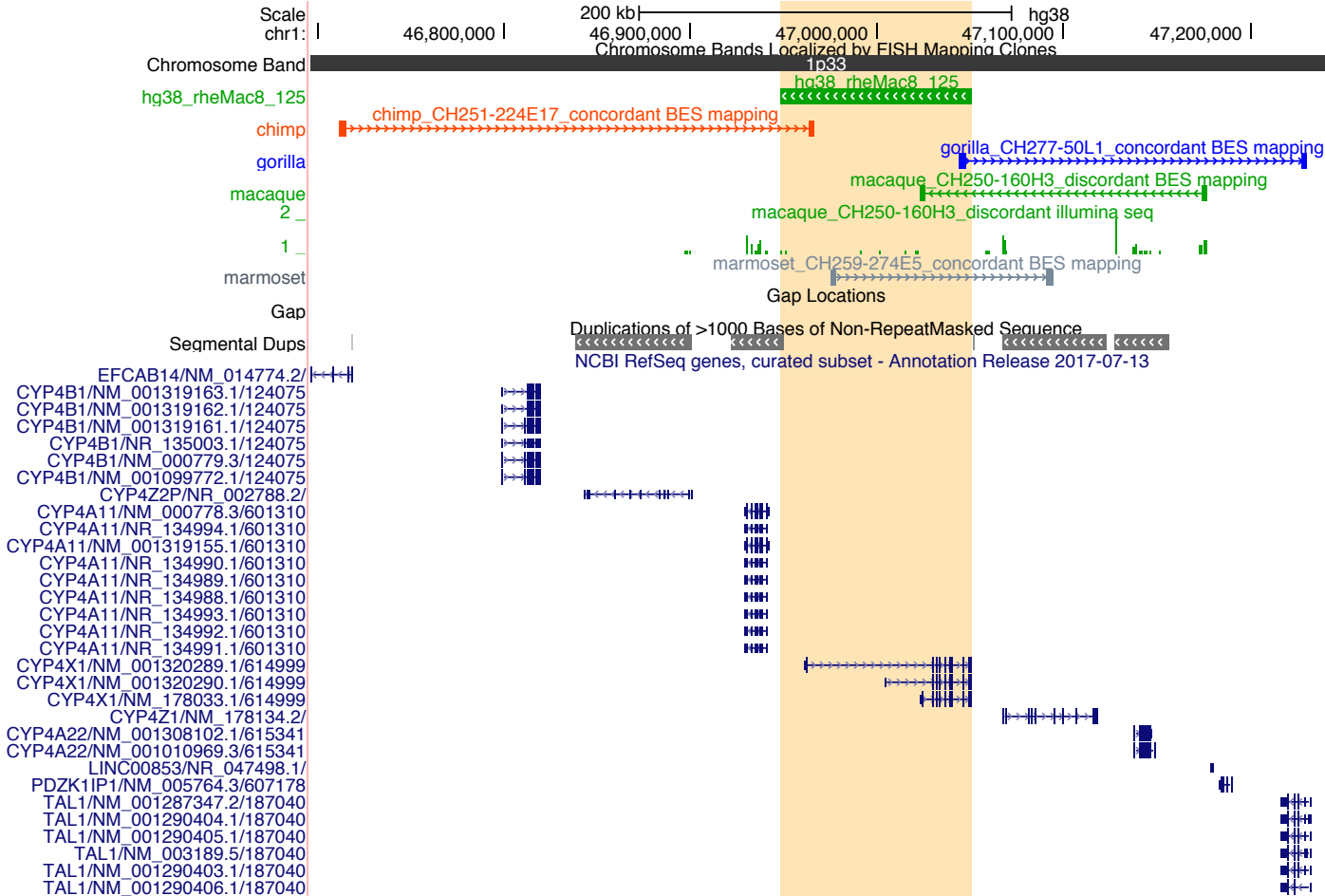
Gap

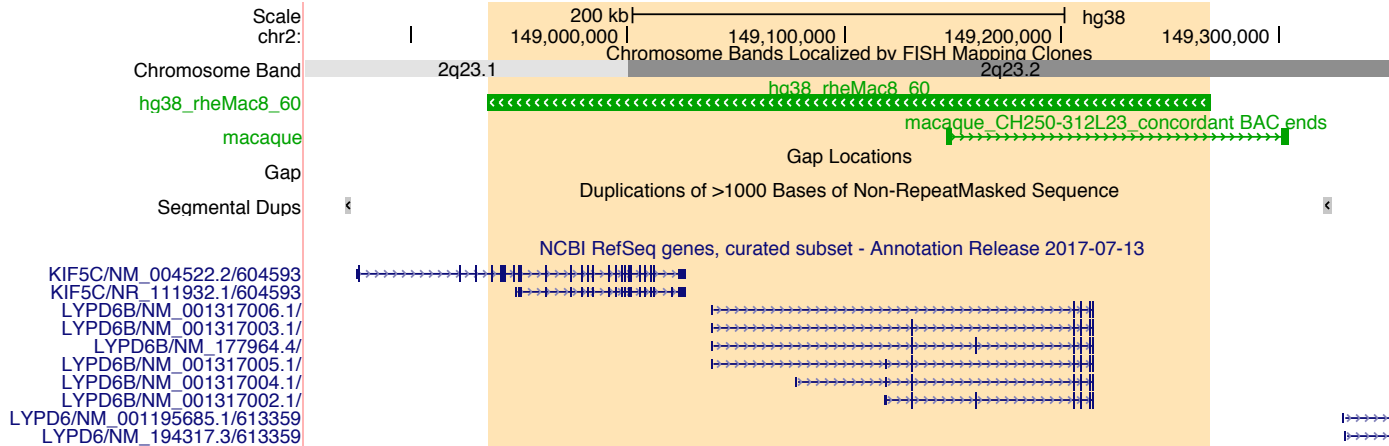
Segmental Dups

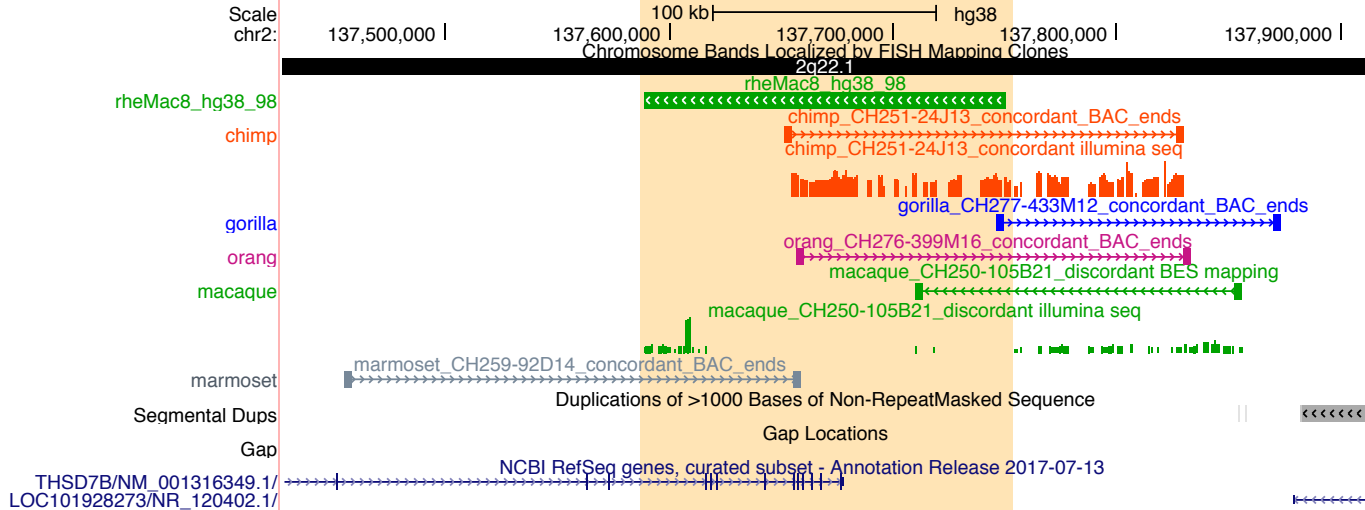
Duplications of >1000 Bases of Non-RepeatMasked Sequence

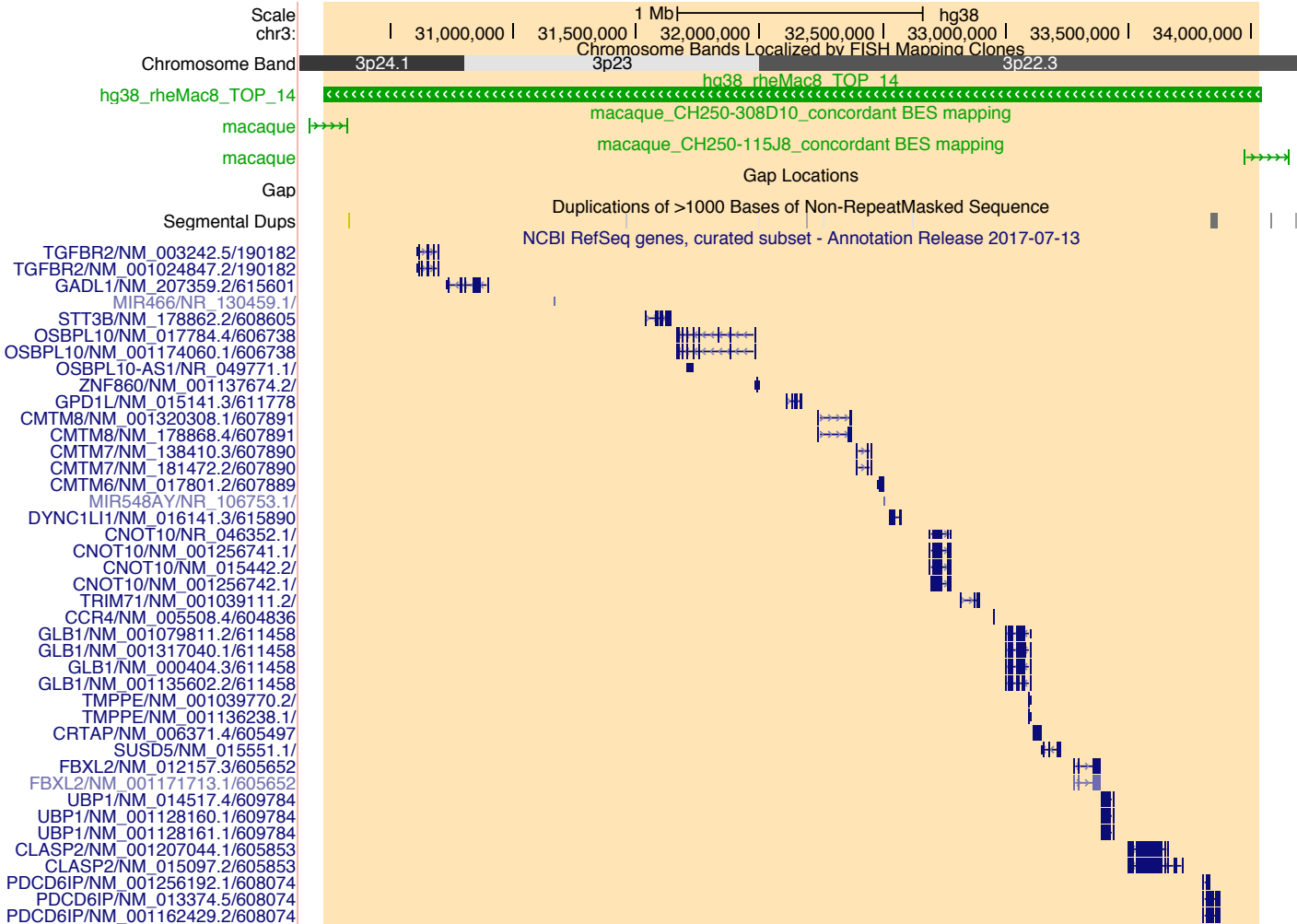
NCBI RefSeq genes, curated subset - Annotation Release 2017-07-13







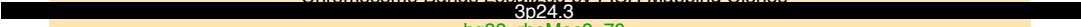




Scale chr3: | 19,500,000 | 20,000,000 | 20,500,000 | 21,000,000 | 21,500,000 | 22,000,000 | 22,500,000 | hg38

Chromosome Bands Localized by FISH Mapping Clones

Chromosome Band



hg38_rheMac8_70



hg38_rheMac8_70

hg38_rheMac8_13



hg38_rheMac8_13

macaque



macaque_CH250-355A8_concordant BES mapping

macaque

macaque_CH250-367M7_concordant BES mapping



Gap

Gap Locations

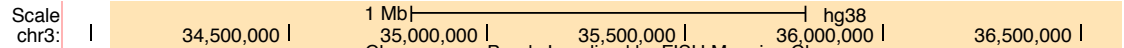
Segmental Dups

Duplications of >1000 Bases of Non-RepeatMasked Sequence

NCBI RefSeq genes, curated subset - Annotation Release 2017-07-13

- KCNH8/NM_144633.2/608260
- MIR4791/NR_039954.1/
- EFHB/NM_144715.3/
- RAB5A/NM_001292048.1/179512
- RAB5A/NM_004162.4/179512
- PP2D1/NR_027694.1/
- PP2D1/NM_001252657.1/
- KAT2B/NM_003884.4/602303
- MIR3135A/NR_036086.1/
- SGO1/NM_001199252.2/609168
- SGO1/NM_001199254.2/609168
- SGO1/NM_001199257.2/609168
- SGO1/NM_001199256.2/609168
- SGO1/NM_001012410.4/609168
- SGO1/NM_001012412.4/609168
- SGO1/NM_138484.4/609168
- SGO1/NM_001199251.2/609168
- SGO1/NM_001199253.2/609168
- SGO1/NM_001199255.2/609168
- SGO1/NM_001012409.3/609168
- SGO1/NM_001012411.3/609168
- SGO1/NM_001012413.3/609168
- SGO1/NR_131179.1/609168
- SGO1/NR_131180.1/609168
- SGO1-AS1/NR_132785.1/
- LOC101927829/NR_110814.1/
- VENTXP7/NR_002311.1/
- ZNF385D/NM_024697.2/
- ZNF385D-AS1/NR_046731.1/
- ZNF385D-AS2/NR_046876.1/





Chromosome Band **3p22.3** **3p22.2**



macaque **macaque_CH250-115J8_concordant BES mapping**



Gap Locations

Gap

Duplications of >1000 Bases of Non-RepeatMasked Sequence

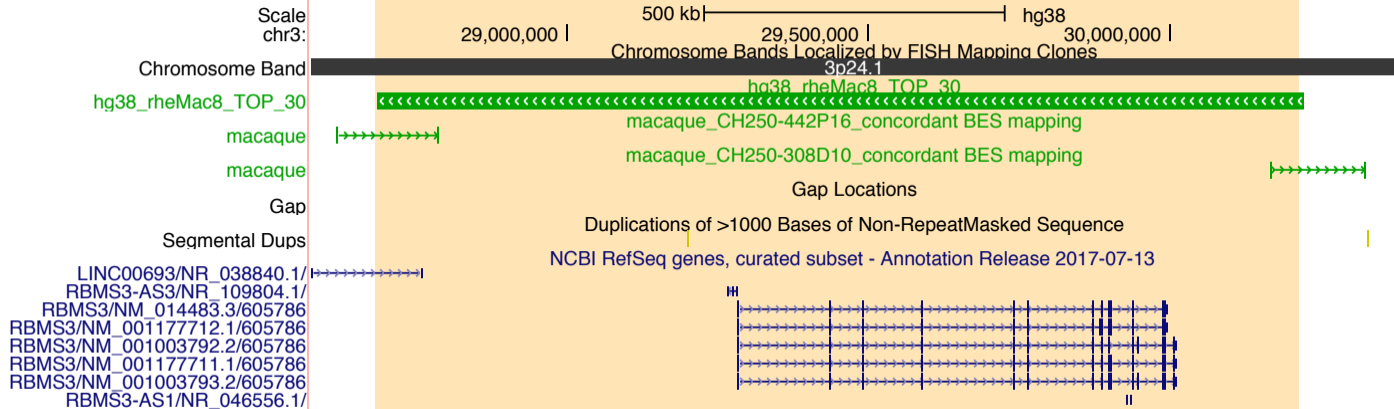
Segmental Dups



NCBI RefSeq genes, curated subset - Annotation Release 2017-07-13

- LOC101928135/NR_110817.1/
- ARPP21/NM_001267616.1/605488
- ARPP21/NM_001267617.1/605488
- ARPP21/NM_016300.4/605488
- ARPP21/NM_001267618.1/605488
- ARPP21/NM_001025068.1/605488
- ARPP21/NM_001025069.1/605488
- ARPP21/NM_001267619.1/605488
- ARPP21/NM_198399.1/605488
- MIR128-2/NR_029824.1/611769
- STAC/NM_001292049.1/602317
- STAC/NM_003149.2/602317





Scale chr4: | 105,000,000 | 105,500,000 | 106,000,000 | 106,500,000 | 107,000,000 | 107,500,000 | 1 Mb hg38

Chromosome Band 4q24 4q25

hg38_rheMac8_14 macaque_CH250-450K19_concordant BES mapping

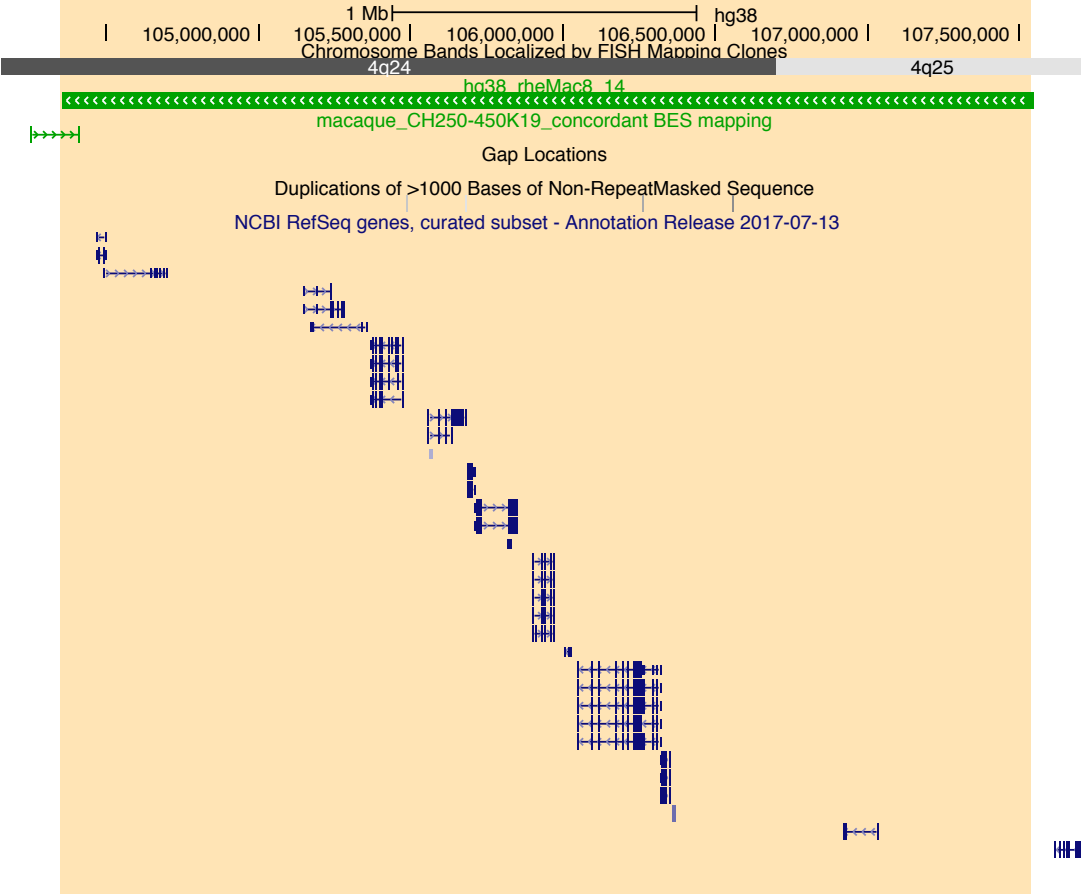
macaque >>>>

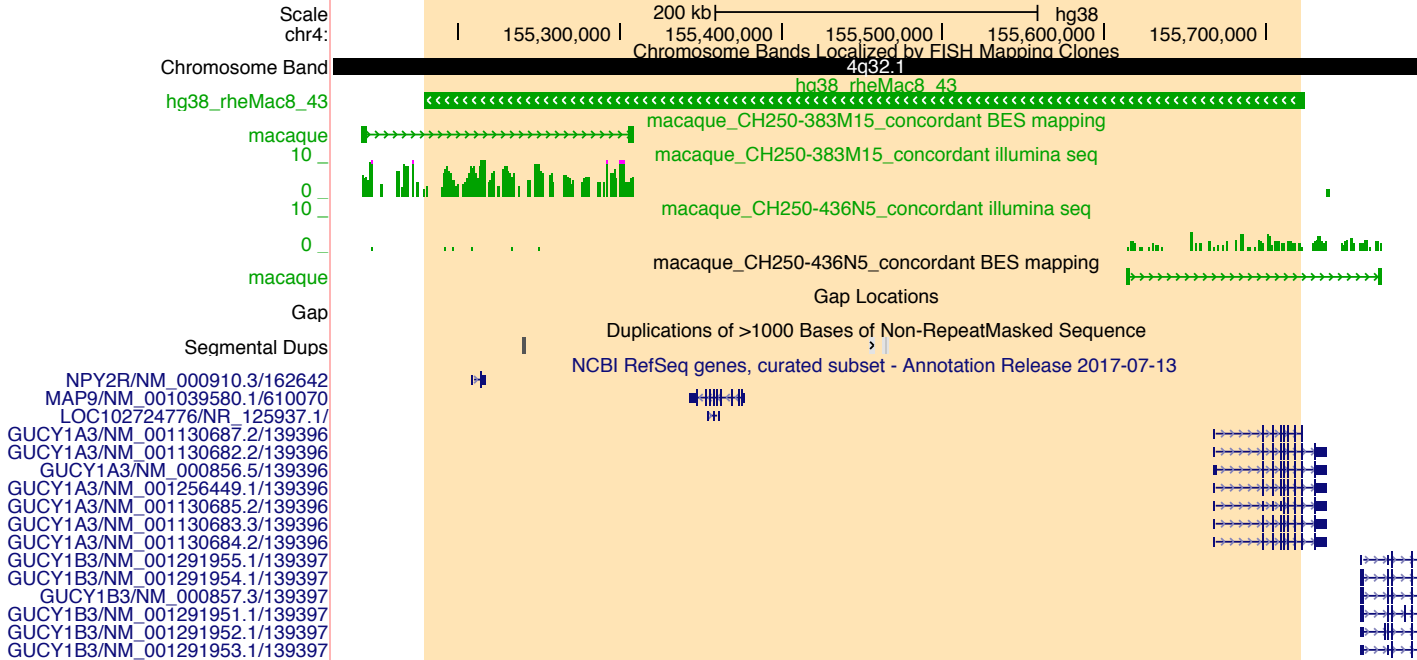
Gap Locations

Duplications of >1000 Bases of Non-RepeatMasked Sequence

NCBI RefSeq genes, curated subset - Annotation Release 2017-07-13

- CXXC4/NR_132741.1/611645
- CXXC4/NM_025212.3/611645
- LOC101929468/NR_125926.1/
- TET2/NM_017628.4/612839
- TET2/NM_001127208.2/612839
- TET2-AS1/NR_126420.1/
- PPA2/NM_176869.2/609988
- PPA2/NM_006903.4/609988
- PPA2/NM_176866.2/609988
- PPA2/NM_176867.3/609988
- ARHGEF38/NM_001242729.1/
- ARHGEF38/NM_017700.1/
- ARHGEF38-IT1/NR_046840.1/
- INTS12/NM_020395.3/611355
- INTS12/NM_001142471.1/611355
- GSTCD/NM_024751.3/615912
- GSTCD/NM_001031720.3/615912
- LOC101929529/NR_125927.1/
- NPNT/NM_001184692.1/610306
- NPNT/NM_001033047.2/610306
- NPNT/NM_001184693.1/610306
- NPNT/NM_001184691.1/610306
- NPNT/NM_001184690.1/610306
- LOC101929577/NR_125928.1/
- TBCK/NM_001290768.1/616899
- TBCK/NM_001163435.2/616899
- TBCK/NM_001163437.2/616899
- TBCK/NM_033115.4/616899
- TBCK/NM_001163436.2/616899
- AIMP1/NM_001142415.1/603605
- AIMP1/NM_004757.3/603605
- AIMP1/NM_001142416.1/603605
- GIMD1/NM_001195138.1/
- DKK2/NM_014421.2/605415
- PAPSS1/NM_005443.4/603262





Scale
chr4:

80,900,000 |

100 kb |

81,000,000 |

81,100,000 |

hg38

81,200,000 |

Chromosome Bands Localized by FISH Mapping Clones

Chromosome Band

4q21.21

hg38_rheMac8_59

hg38_rheMac8_59

macaque

macaque_CH250-399L19_concordant BES mapping

macaque

macaque_CH250-326I9_concordant BES mapping

Gap

Gap Locations

Duplications of >1000 Bases of Non-RepeatMasked Sequence

Segmental Dups

NCBI RefSeq genes, curated subset - Annotation Release 2017-07-13

C4orf22/NM_152770.2/

C4orf22/NM_001206997.1/

BMP3/NM_001201.2/112263

PRKG2/NM_001282480.1/601591

PRKG2/NM_001282481.1/601591

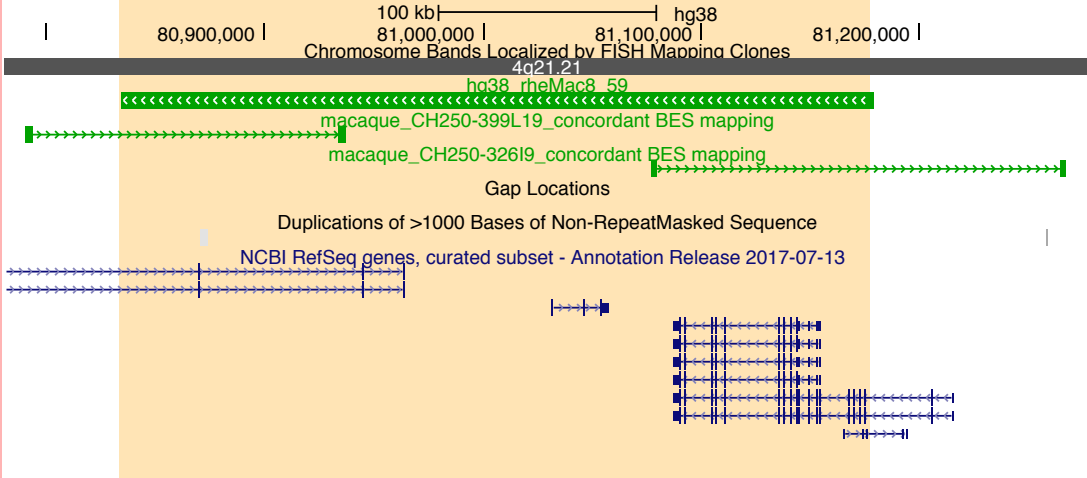
PRKG2/NM_001282483.1/601591

PRKG2/NM_001282482.1/601591

PRKG2/NM_006259.2/601591

PRKG2/NM_001282485.1/601591

LOC101928942/NR_125908.1/



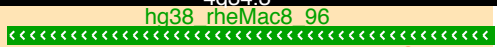
Scale
chr4:

178,200,000 | 100 kb | 178,300,000 | 178,400,000 | hg38

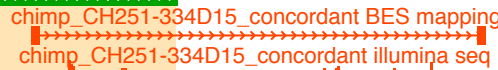
Chromosome Bands Localized by FISH Mapping Clones

4q34.3

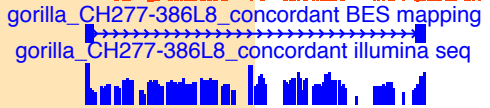
hg38_rheMac8_96



chimp



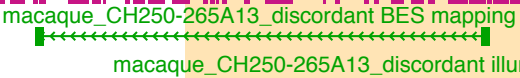
gorilla



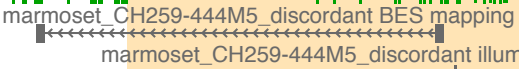
orang



macaque



marmoset



Gap

Gap Locations

Segmental Dups

Duplications of >1000 Bases of Non-RepeatMasked Sequence

NCBI RefSeq genes, curated subset - Annotation Release 2017-07-13

Scale
chr5:

99,500,000 |

500 kb |

100,000,000 |

hg38

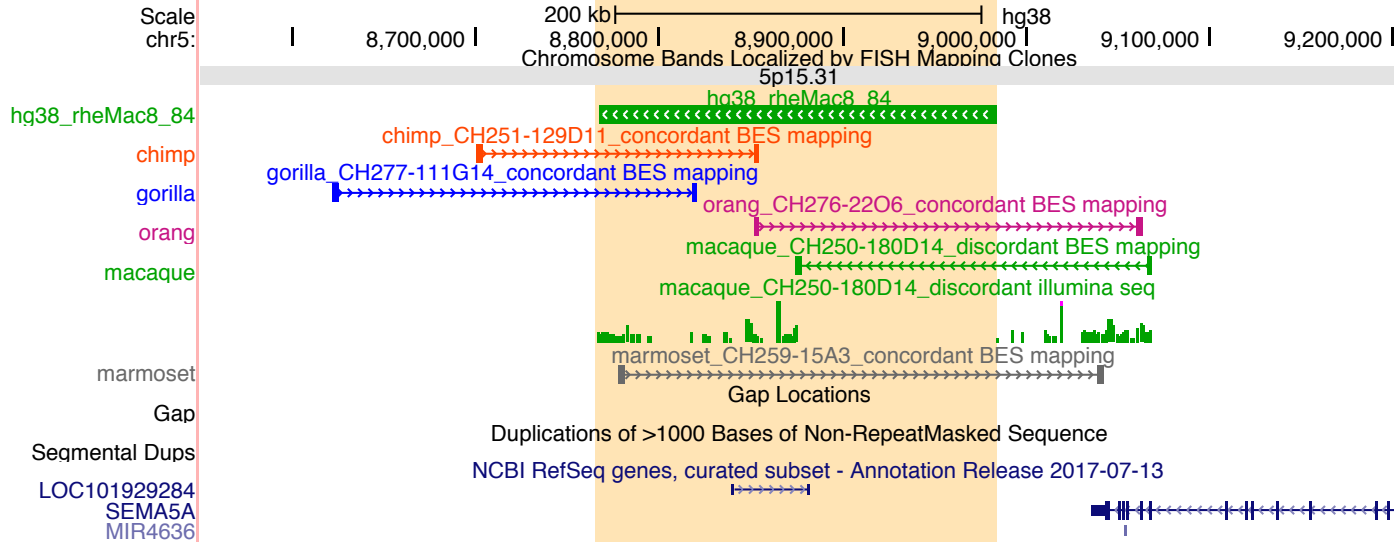
100,500,000 |

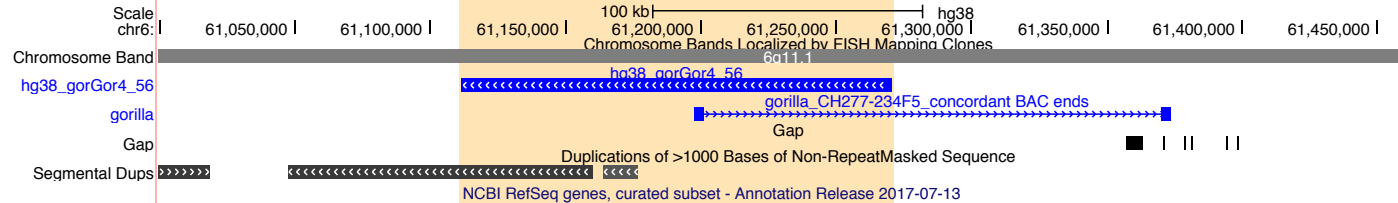
Chromosome Bands I localized by FISH Mapping Clones

5q21.1

hg38_panTro5_60
hg38_panTro5_38
hg38_ponAbe2_24
hg38_rheMac8_32
chimp
chimp
gorilla
orang
macaque
macaque
Gap
Segmental Dups
CTD-2151A2.1
LOC100133050
FAM174A

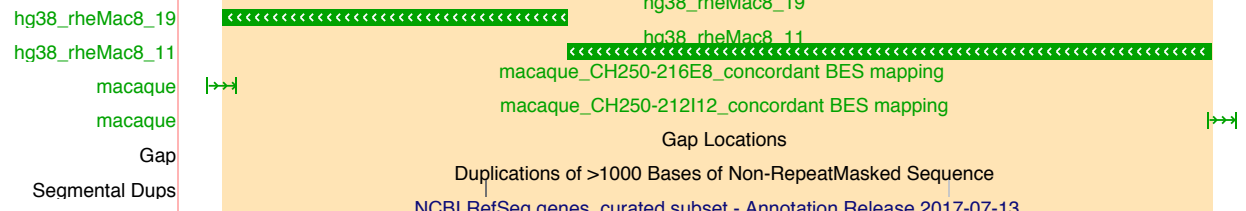




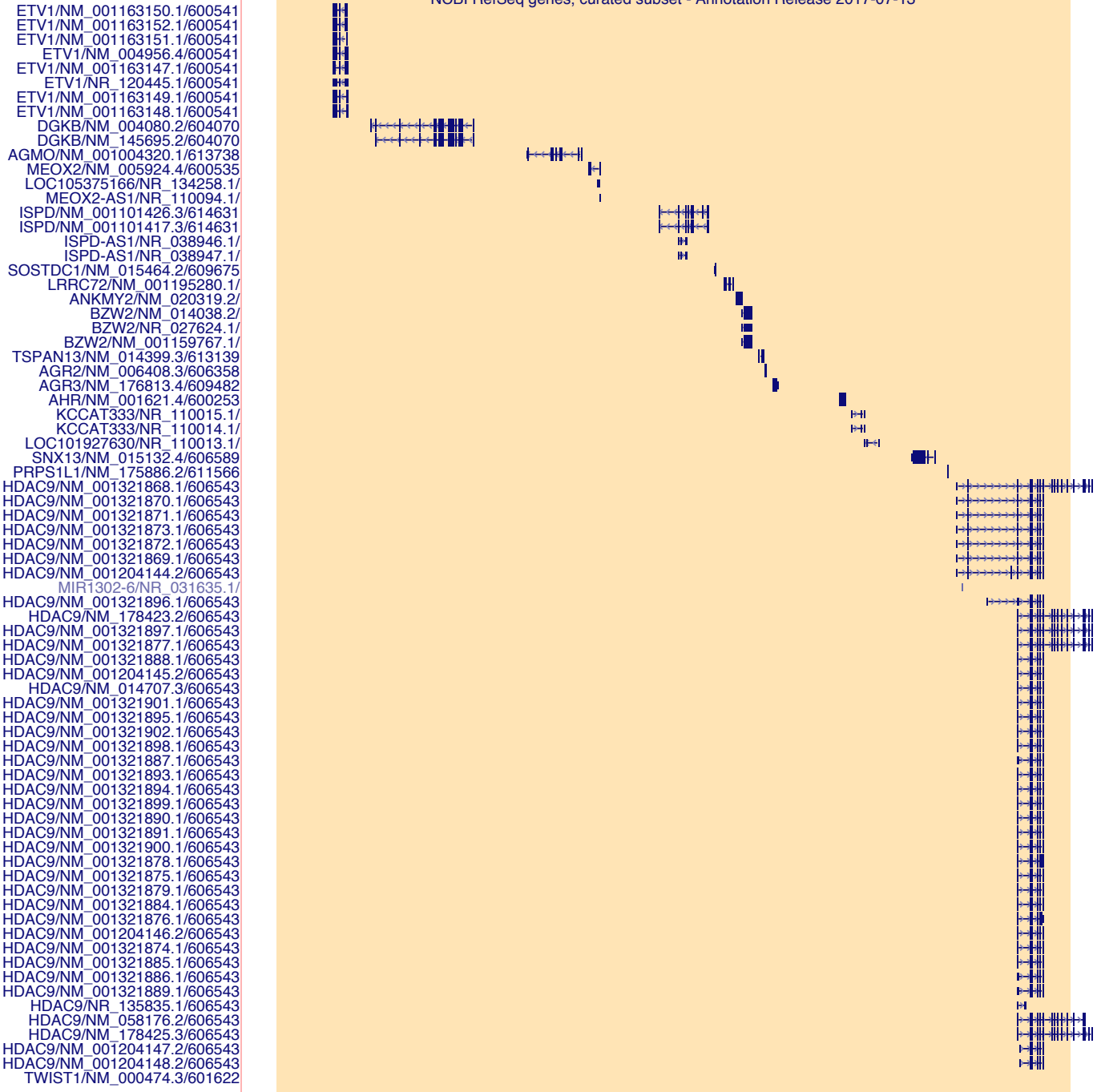


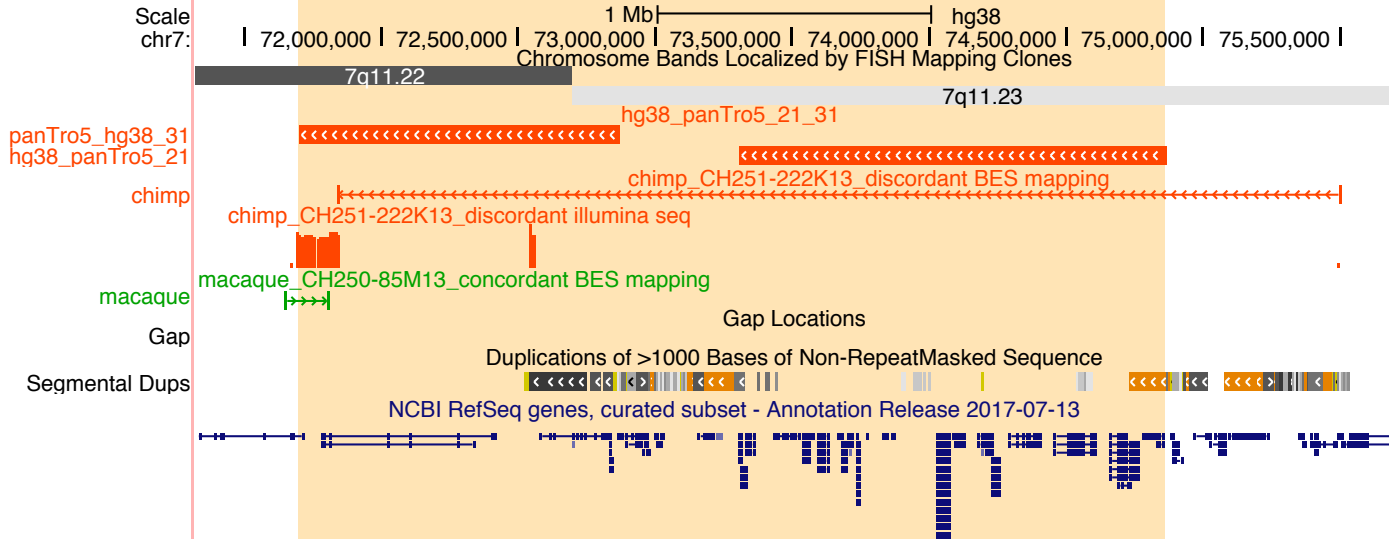
Scale chr7: 14,000,000 | 15,000,000 | 16,000,000 | 17,000,000 | 18,000,000 | 19,000,000 | hg38
2 Mb | FISH Mapping Clones

Chromosome Band 7p21.3 | 7p21.2 | 7p21.1



Duplications of >1000 Bases of Non-RepeatMasked Sequence
NCBI RefSeq genes, curated subset - Annotation Release 2017-07-13





Scale chr22: 32,100,000 | 100 kb | 32,150,000 | 32,200,000 | hg38 32,250,000 |

Chromosome Band 22q12.3

hg38_rheMac8_118

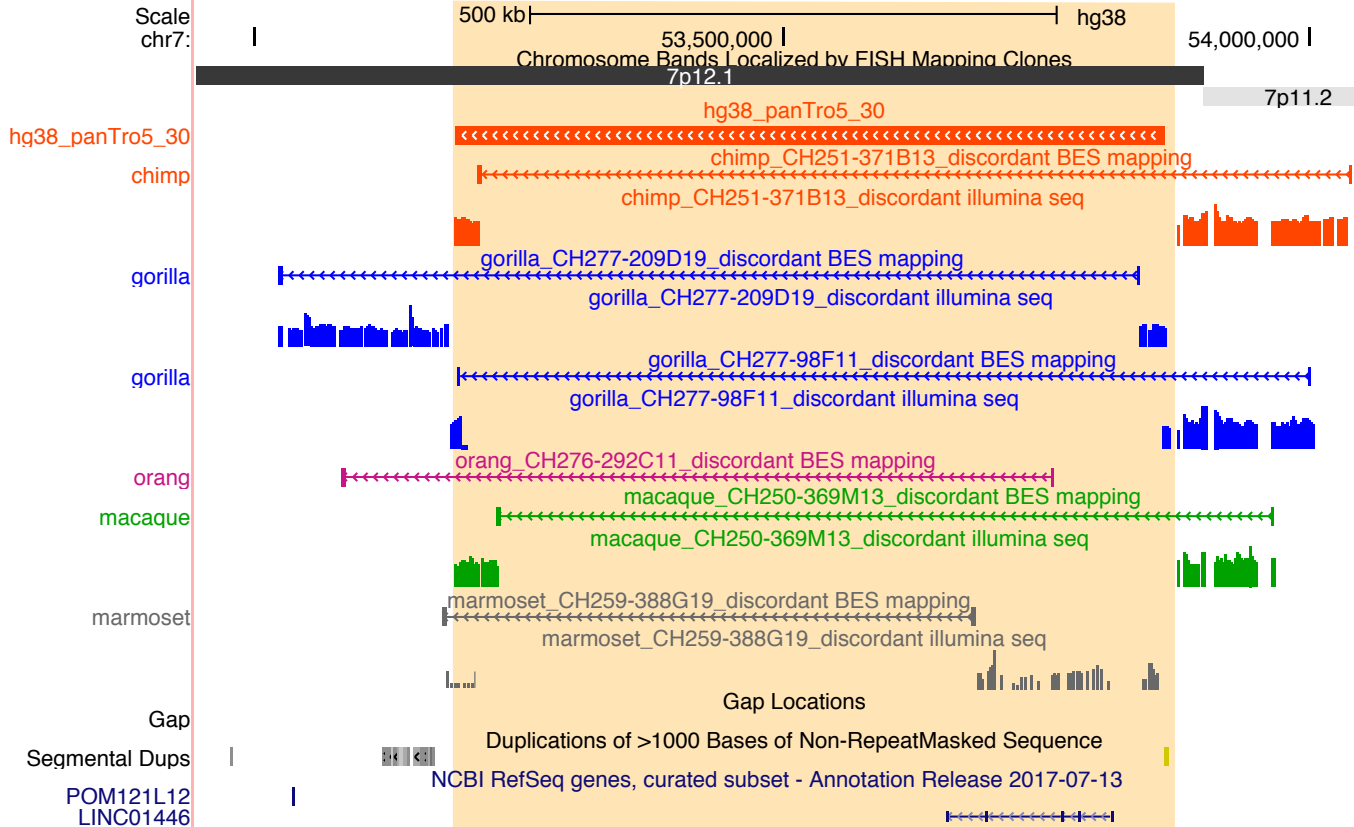
macaque macaque CH250-530L10 concordant BES mapping

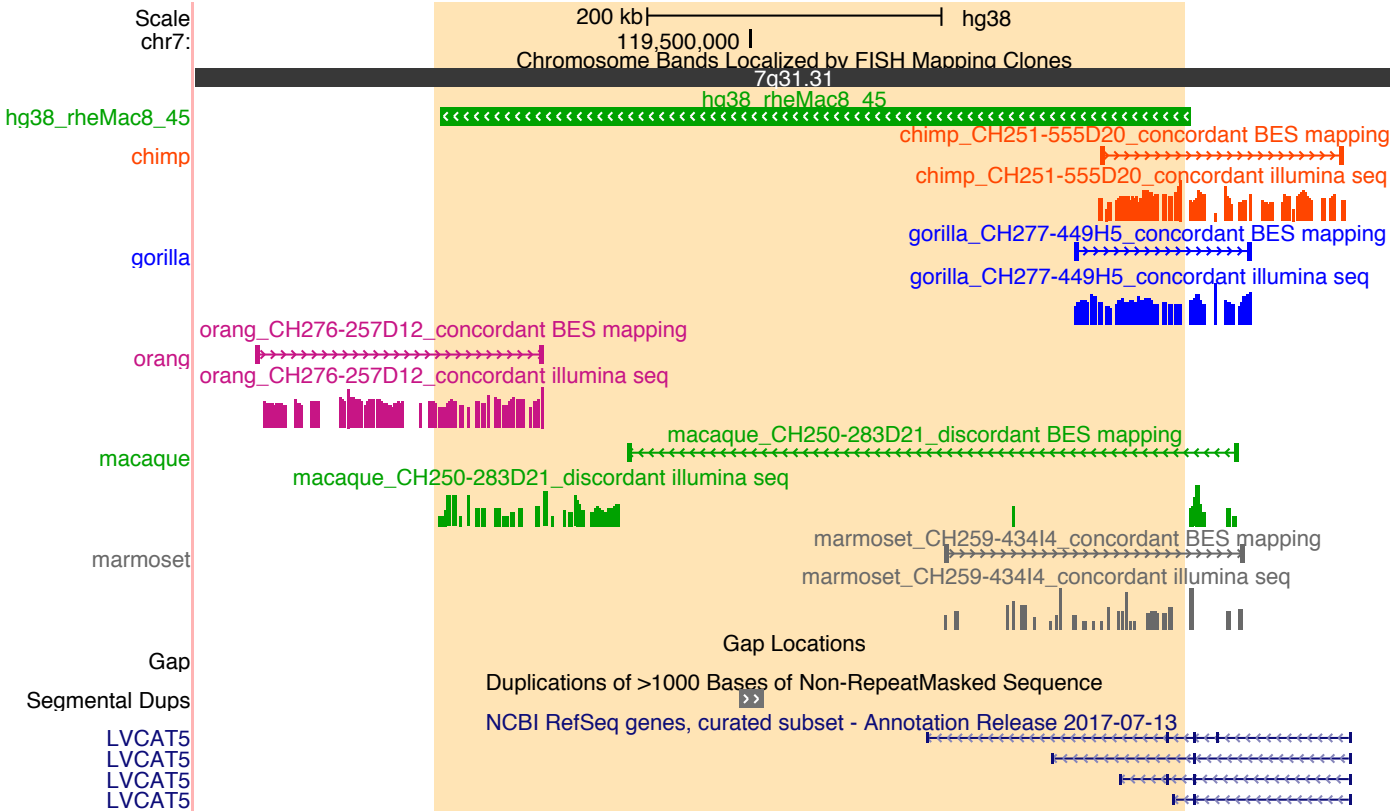
Gap Gap Locations

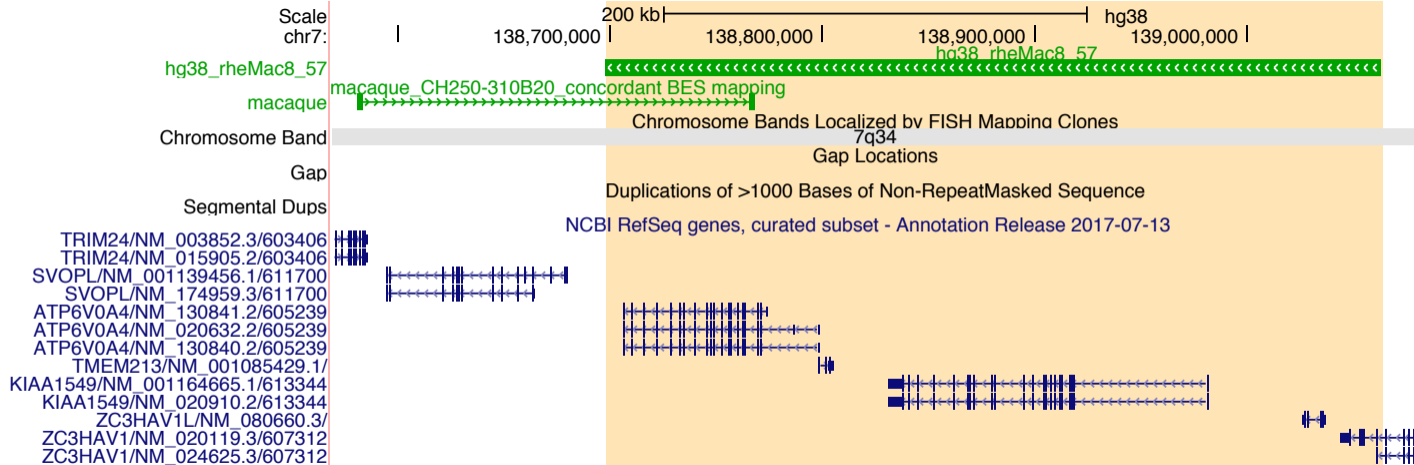
Segmental Dups Duplications of >1000 Bases of Non-RepeatMasked Sequence

NCBI RefSeq genes, curated subset - Annotation Release 2017-07-13

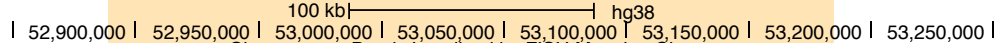








Scale
chr7:

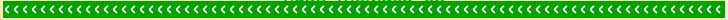


Chromosome Bands Localized by FISH Mapping Clones

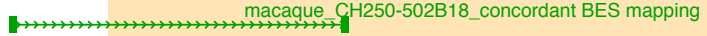
Chromosome Band



hg38_rheMac8_68



macaque



macaque



Gap

Gap Locations

Segmental Dups

Duplications of >1000 Bases of Non-RepeatMasked Sequence



NCBI RefSeq genes, curated subset - Annotation Release 2017-07-13

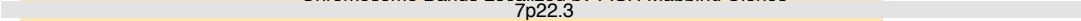
POM121L12/NM_182595.3/



Scale chr7: | 850,000 | 100 kb | 900,000 | 950,000 | 1,000,000 | hg38 | 1,050,000 |

Chromosome Bands Localized by FISH Mapping Clones

Chromosome Band



hg38 panTro5 TOP_36

hg38_panTro5_TOP_36



chimp_CH251-297P2_concordant BES mapping

chimp



Gap Locations

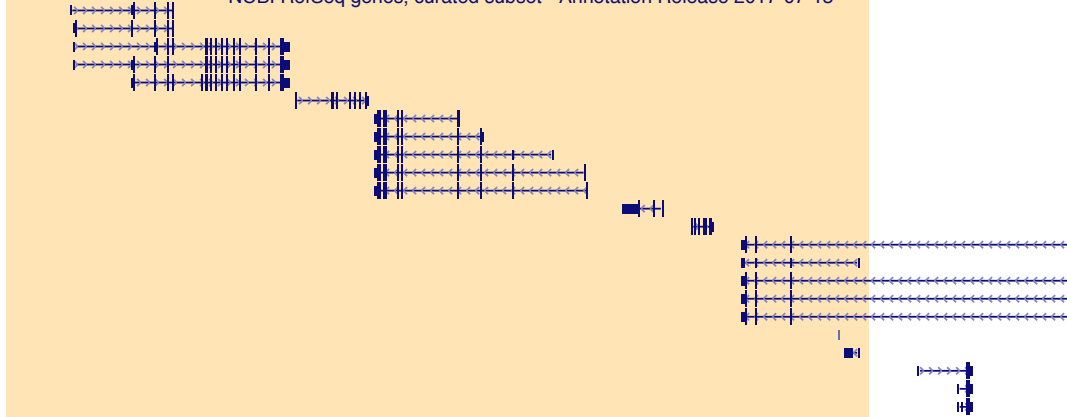
Gap

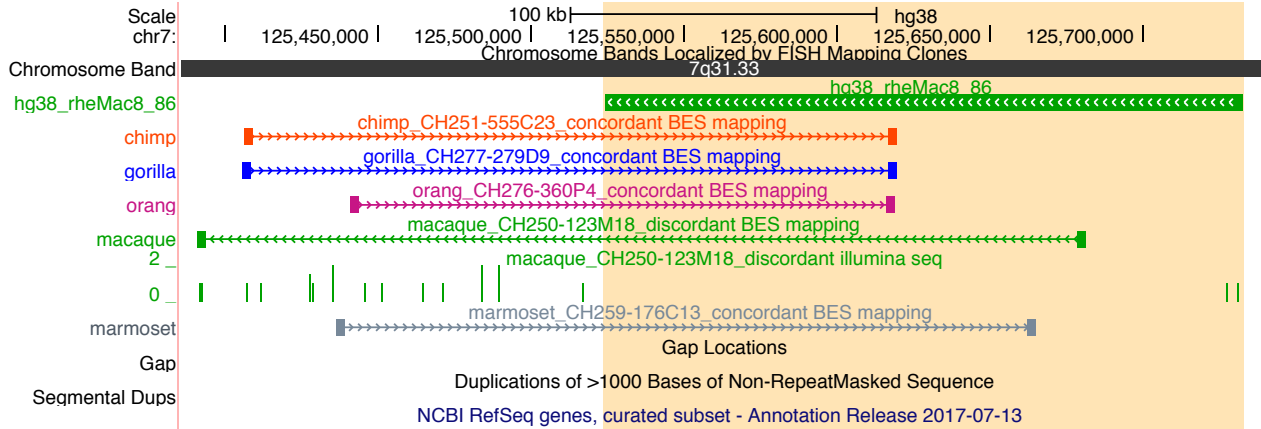
Duplications of >1000 Bases of Non-RepeatMasked Sequence

Segmental Dups

NCBI RefSeq genes, curated subset - Annotation Release 2017-07-13

- SUN1/NM_001171946.1/607723
- SUN1/NM_001171945.1/607723
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- SUN1/NM_001171944.1/607723
- SUN1/NM_001130965.2/607723
- GET4/NM_015949.2/612056
- ADAP1/NM_001284311.1/608114
- ADAP1/NM_001284310.1/608114
- ADAP1/NM_001284309.1/608114
- ADAP1/NM_006869.3/608114
- ADAP1/NM_001284308.1/608114
- COX19/NM_001031617.2/610429
- CYP2W1/NM_017781.2/615967
- C7orf50/NM_001318252.1/
- C7orf50/NR_134537.1/
- C7orf50/NM_032350.5/
- C7orf50/NM_001134395.1/
- C7orf50/NM_001134396.1/
- MIR339/NR_029898.1/615977
- C7orf50/NR_134538.1/
- GPR146/NM_001303473.1/
- GPR146/NM_138445.3/
- GPR146/NM_001303474.1/





Scale chr7: | 2,450,000 | 2,500,000 | 2,550,000 | 2,600,000 | 2,650,000 | 2,700,000 | 2,750,000 | 2,800,000 | hg38

Chromosome Bands Localized by FISH Mapping Clones
7p22.3

hg38_rheMac8_101



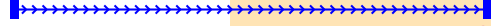
chimp_CH251-294P16_concordant BES mapping

chimp



gorilla

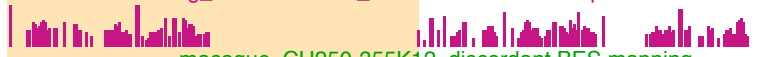
gorilla_CH277-46C23_concordant BES mapping



orang

orang_CH276-266M8_discordant BES mapping

orang_CH276-266M8_discordant illumina seq



macaque

macaque_CH250-355K12_discordant BES mapping

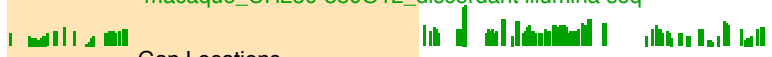
macaque_CH250-355K12_discordant illumina seq



macaque

macaque_CH250-380O12_discordant BES mapping

macaque_CH250-380O12_discordant illumina seq



Gap Locations

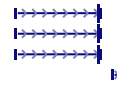
Gap

Duplications of >1000 Bases of Non-RepeatMasked Sequence

Segmental Dups

NCBI RefSeq genes, curated subset - Annotation Release 2017-07-13

CHST12
CHST12
CHST12
LOC101927181



GRIFIN

LFNG

IQCE

AMZ1

AMZ1

LFNG

IQCE

AMZ1

LFNG

IQCE

AMZ1

MIR46481

IQCE

AMZ1

GNA12

GNA12

GNA12

GNA12

BRAT1

TTYH3



Scale chr8: | 51,000,000 | 51,500,000 | 52,000,000 | 52,500,000 | 53,000,000 | 53,500,000 | hg38

Chromosome Bands 8q11.21 8q11.22 8q11.23

hg38_rheMac8_12

macaque macaque_CH250-400A18_concordant BES mapping

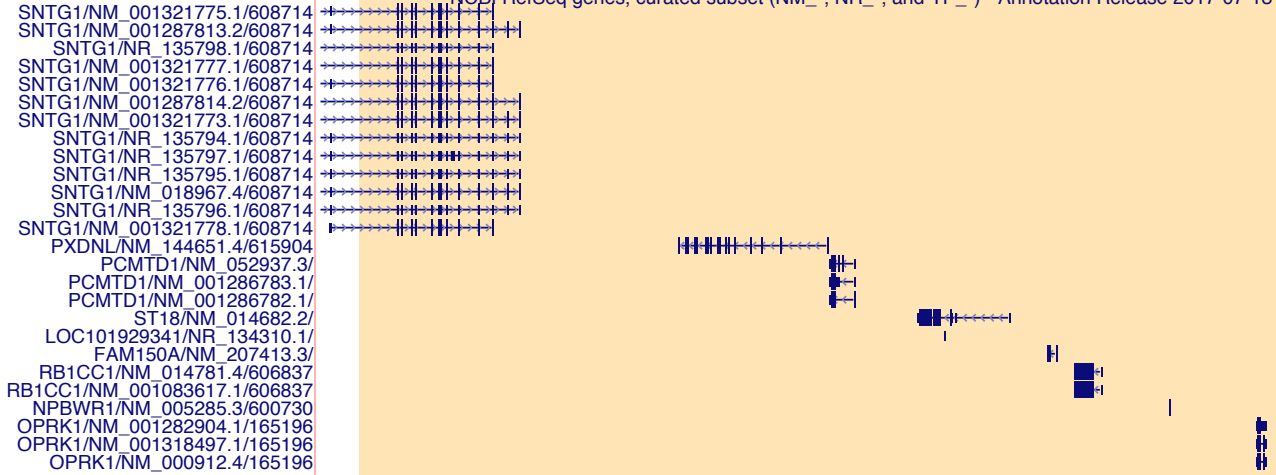
macaque macaque_CH250-426N6_concordant BES mapping

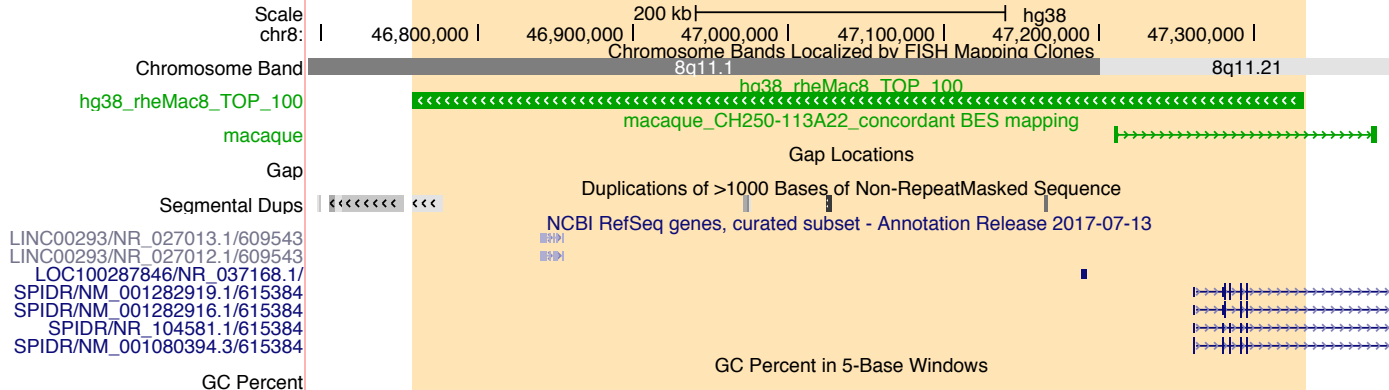
Gap Gap Locations

Duplications of >1000 Bases of Non-RepeatMasked Sequence

Segmental Dups

NCBI RefSeq genes, curated subset (NM_*, NR_*, and YP_*) - Annotation Release 2017-07-13





Scale chr9: | 24,350,000 | 24,400,000 | 24,450,000 | 24,500,000 | 24,550,000 | 24,600,000 | 24,650,000 | 24,700,000 | hg38

Chromosome Bands Localized by FISH Mapping Clones

9p21.3

hg38_rheMac8_107



chimp



gorilla



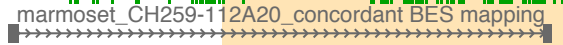
orang



macaque



marmoset



Gap

Gap Locations

Segmental Dups

Duplications of >1000 Bases of Non-RepeatMasked Sequence

IZUMO3

NCBI RefSeq genes, curated subset - Annotation Release 2017-07-13



Scale chr10: | 106,000,000 | 106,500,000 | 107,000,000 | 107,500,000 | hg38

1 Mb

Chromosome Bands Localized by FISH Mapping Clones

Chromosome Band

10q25.1

hg38_rheMac8_16

hg38_rheMac8_16

macaque

macaque_CH250-216G12_concordant BES mapping

macaque

macaque_CH250-437D12_concordant BES mapping

Gap Locations

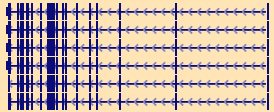
Gap

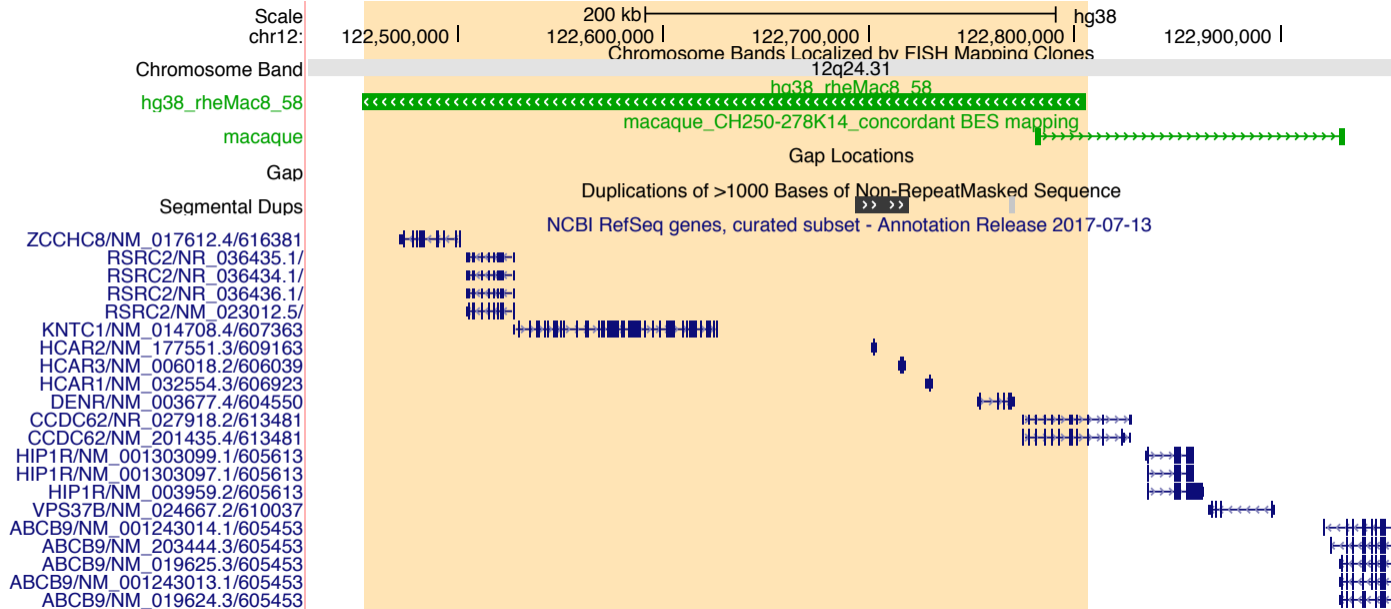
Duplications of >1000 Bases of Non-RepeatMasked Sequence

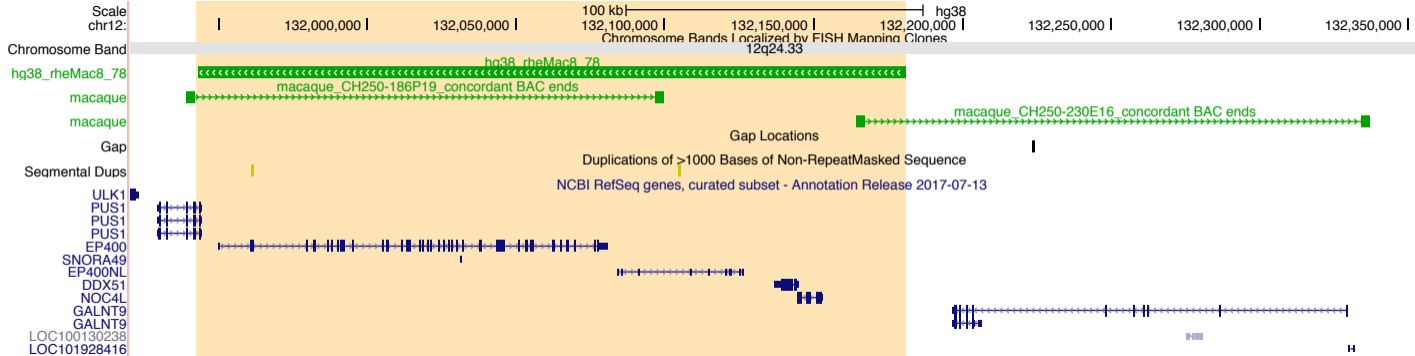
Segmental Dups

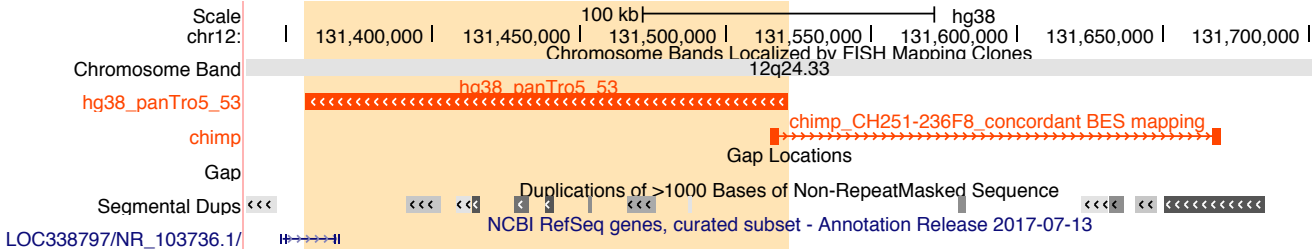
NCBI RefSeq genes, curated subset (NM_*, NR_*, and YP_*) - Annotation Release 2017-07-13

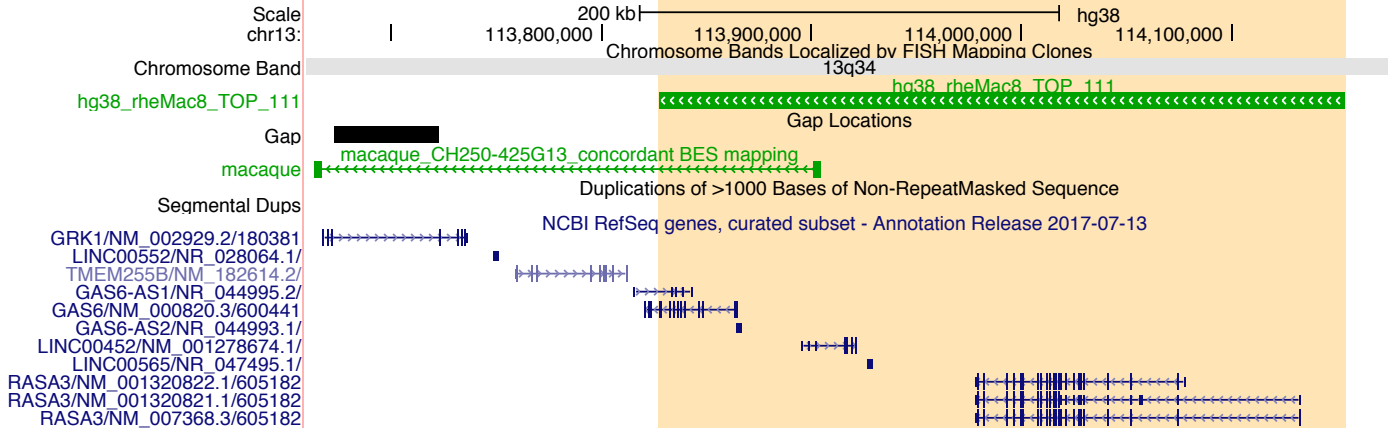
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- LOC105378470/NR_134330.1/
- LOC105378470/NR_134331.1/
- LOC105378470/NR_134329.1/
- SORCS1/NM_001206572.1/606283
- SORCS1/NM_001013031.2/606283
- SORCS1/NM_052918.4/606283
- SORCS1/NM_001206570.1/606283
- SORCS1/NM_001206571.1/606283
- SORCS1/NM_001206569.1/606283
- LINC01435/NR_125760.1/

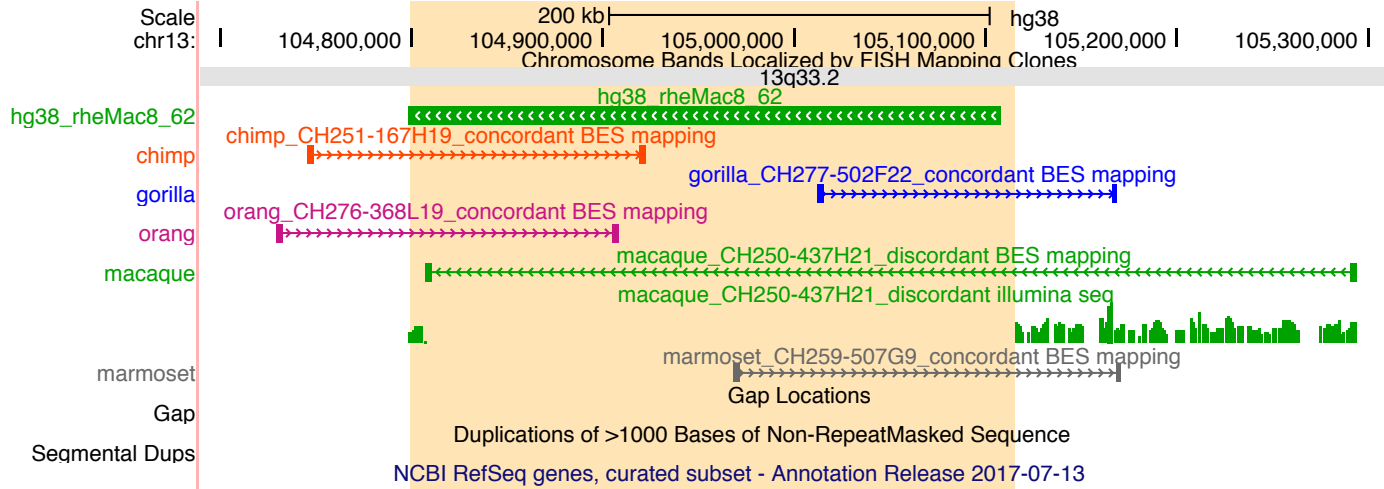


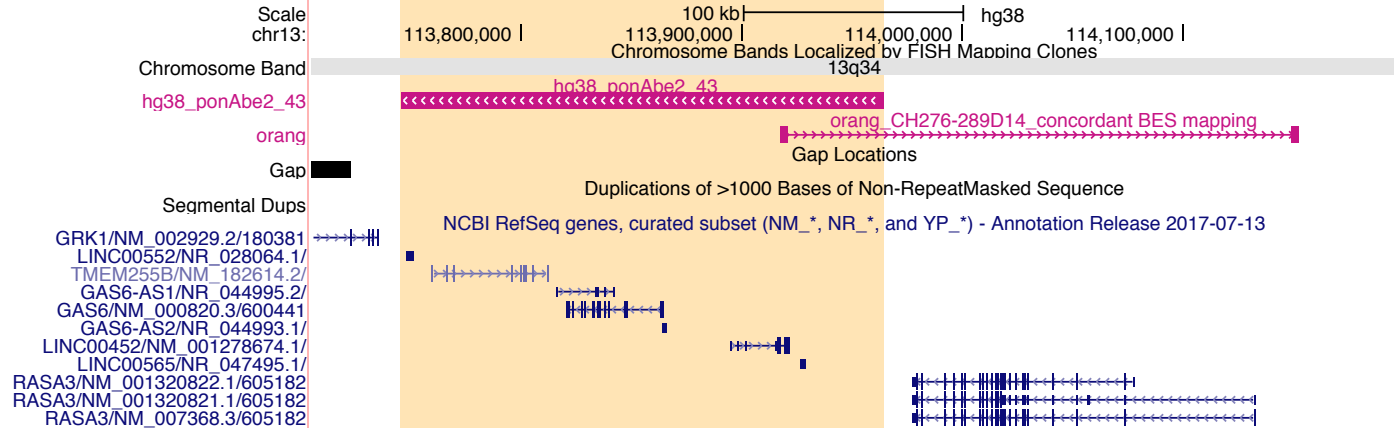












Scale chr14: | 19,800,000 | 19,900,000 | 20,000,000 | 20,100,000 | 20,200,000 | 20,300,000 | hg38

Chromosome Bands Localized by FISH Mapping Clones

14q11.2

hg38_rheMac8_TOP_105



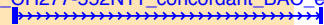
chimp

chimp_CH251-328P13_concordant_BAC_ends



gorilla

gorilla_CH277-552N11_concordant_BAC_ends



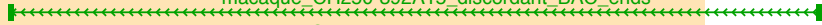
orang

orang_CH276-409H20_concordant_BAC_ends



macaque

macaque_CH250-392A15_discordant_BAC_ends



macaque_CH250-392A15_discordant illumina seq



Segmental Dups



Duplications of >1000 Bases of Non-RepeatMasked Sequence

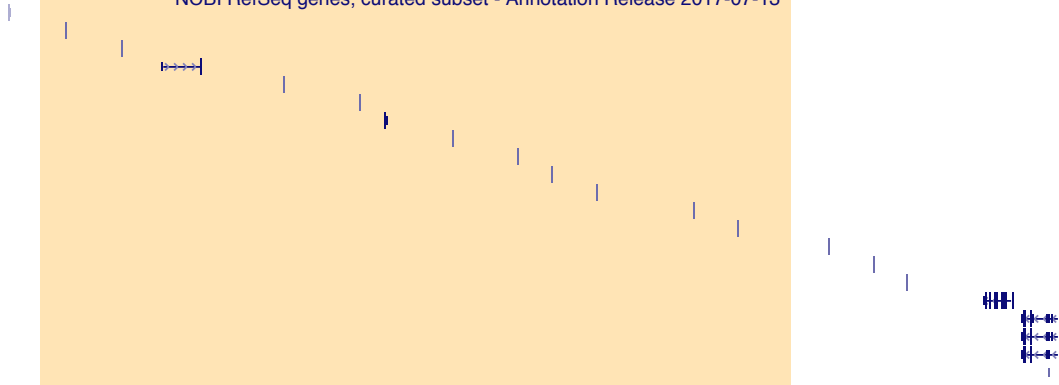


Gap

Gap Locations

NCBI RefSeq genes, curated subset - Annotation Release 2017-07-13

- OR11H2/NM_001197287.1/
- OR4Q3/NM_172194.1/
- OR4M1/NM_001005500.1/
- OR4N2/NM_001004723.2/
- OR4K2/NM_001005501.1/
- OR4K5/NM_001005483.1/
- OR4K1/NM_001004063.2/
- OR4K15/NM_001005486.1/
- OR4K14/NM_001004712.1/
- OR4K13/NM_001004714.1/
- OR4L1/NM_001004717.1/
- OR4K17/NM_001004715.1/
- OR4N5/NM_001004724.1/
- OR11G2/NM_001005503.1/
- OR11H6/NM_001004480.1/
- OR11H4/NM_001004479.1/
- TTC5/NM_138376.2/
- CCNB1IP1/NM_182852.3/608249
- CCNB1IP1/NM_021178.4/608249
- CCNB1IP1/NM_182849.2/608249
- SNORD126/NR_003693.1/



Scale chr15: 26,000,000 | 500 kb | 26,500,000 | hg38 27,000,000 |

Chromosome Band 15q12

hg38_rheMac8_35

hg38_rheMac8_52

macaque macaque_CH250-336A11_concordant BES mapping

Gap Locations

Duplications of >1000 Bases of Non-RepeatMasked Sequence

NCBI RefSeq genes, curated subset - Annotation Release 2017-07-13

ATP10A/NM_024490.3/605855

MIR4715/NR_039865.1/

LOC100128714/NR_040082.1/

LINC00929/NR_038851.1/

LINC00929/NR_038852.1/

GABRB3/NM_001191321.2/137192

GABRB3/NM_001191320.1/137192

GABRB3/NM_001278631.1/137192

GABRB3/NM_000814.5/137192

GABRB3/NM_021912.4/137192

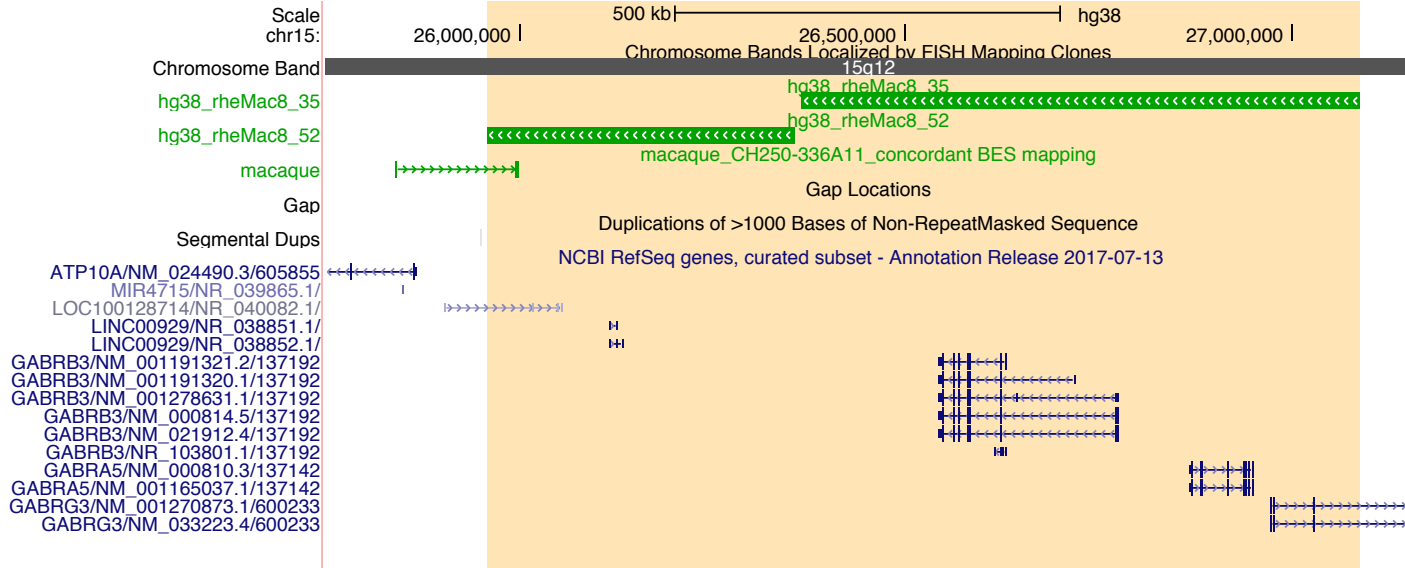
GABRB3/NR_103801.1/137192

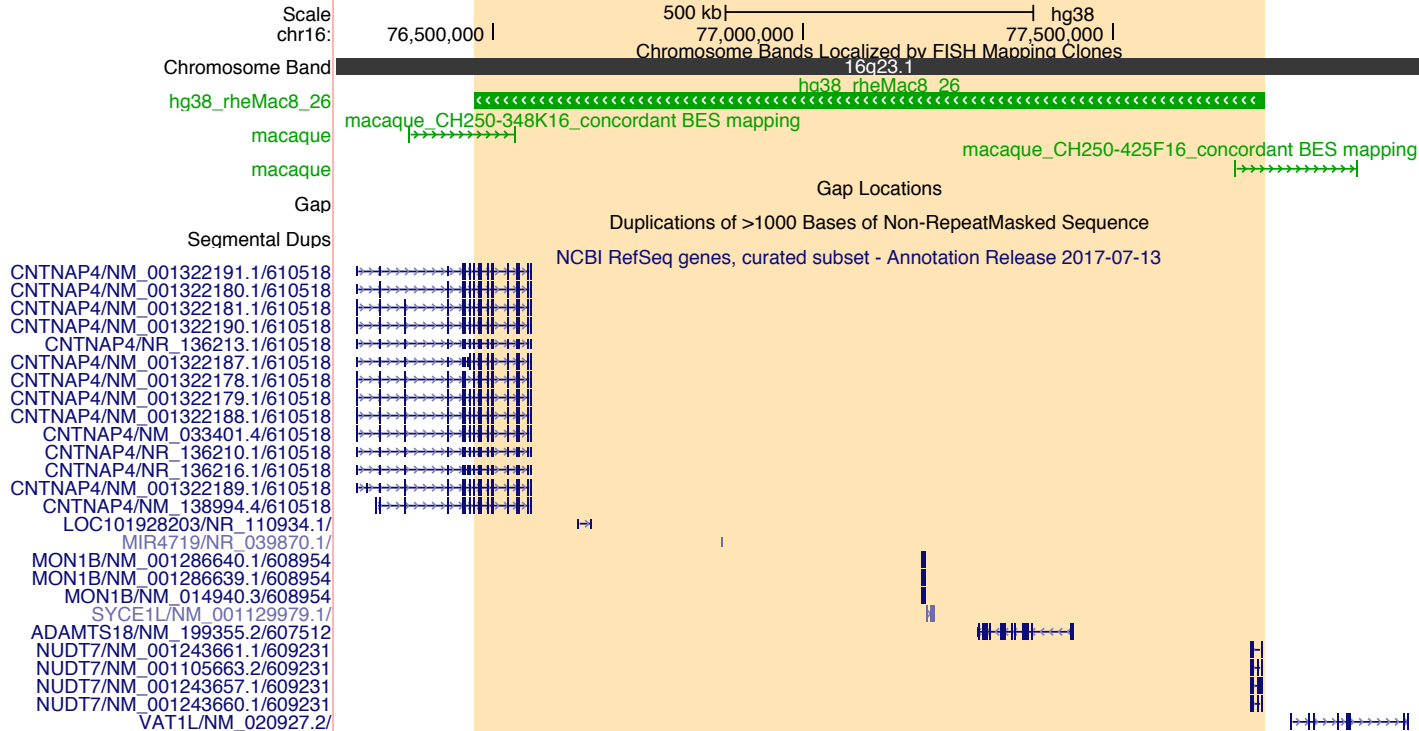
GABRA5/NM_000810.3/137142

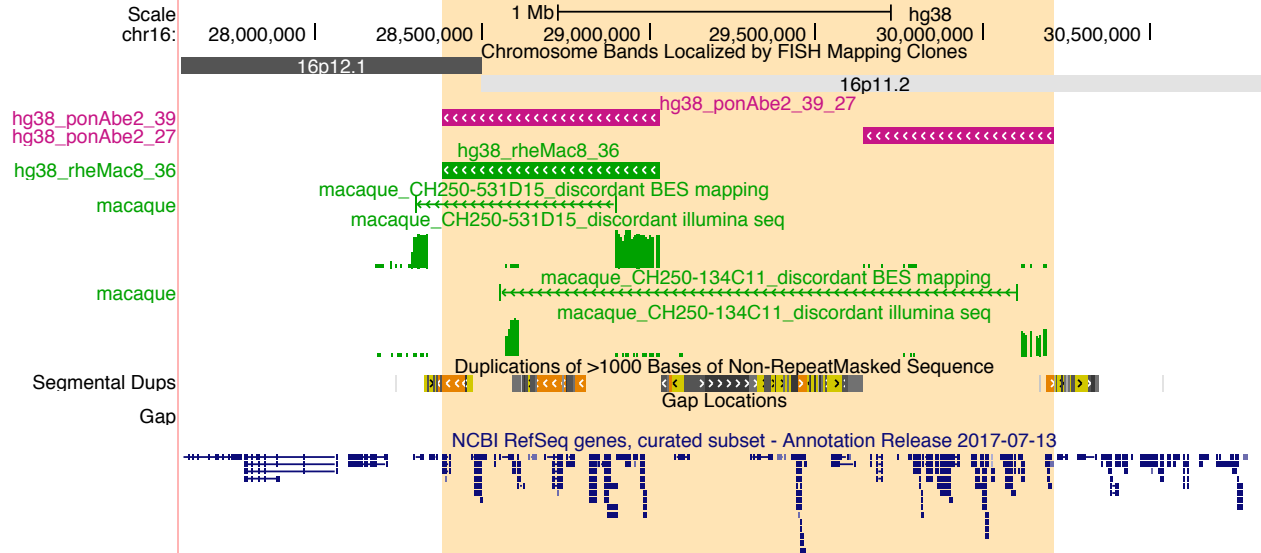
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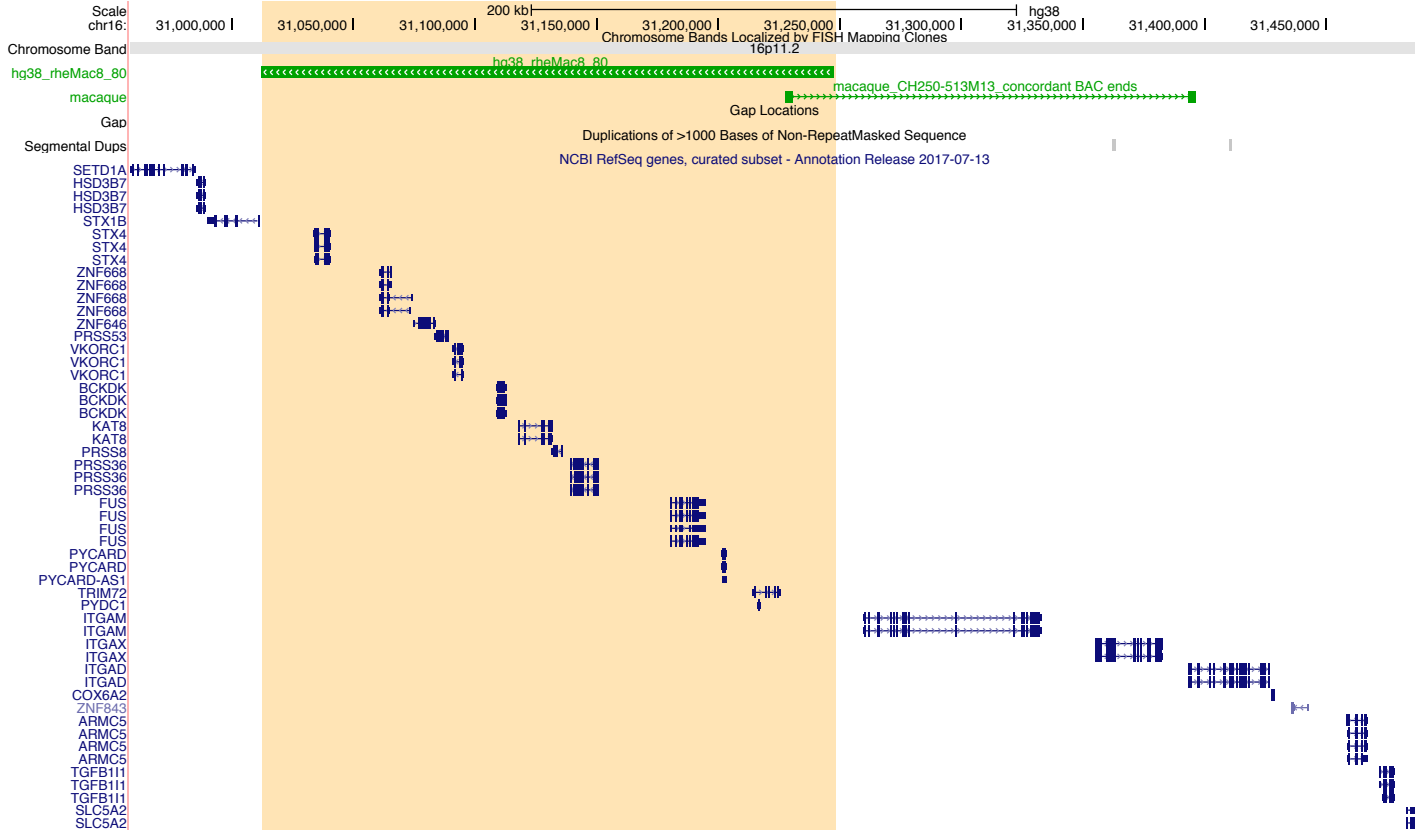
GABRG3/NM_001270873.1/600233

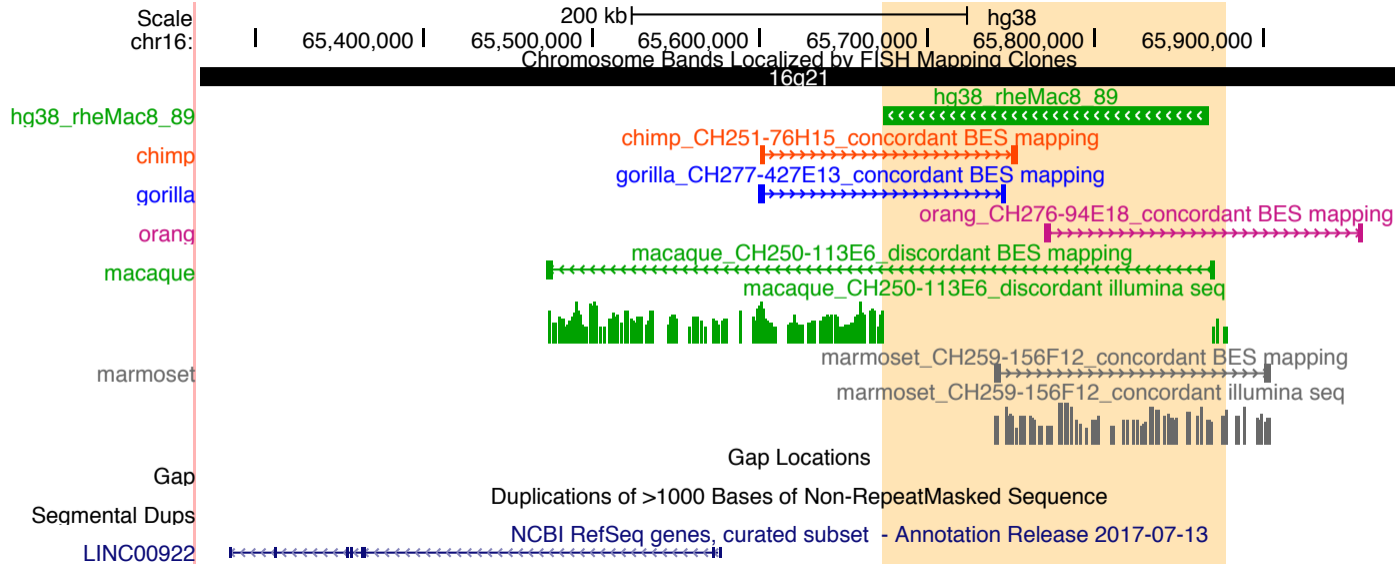
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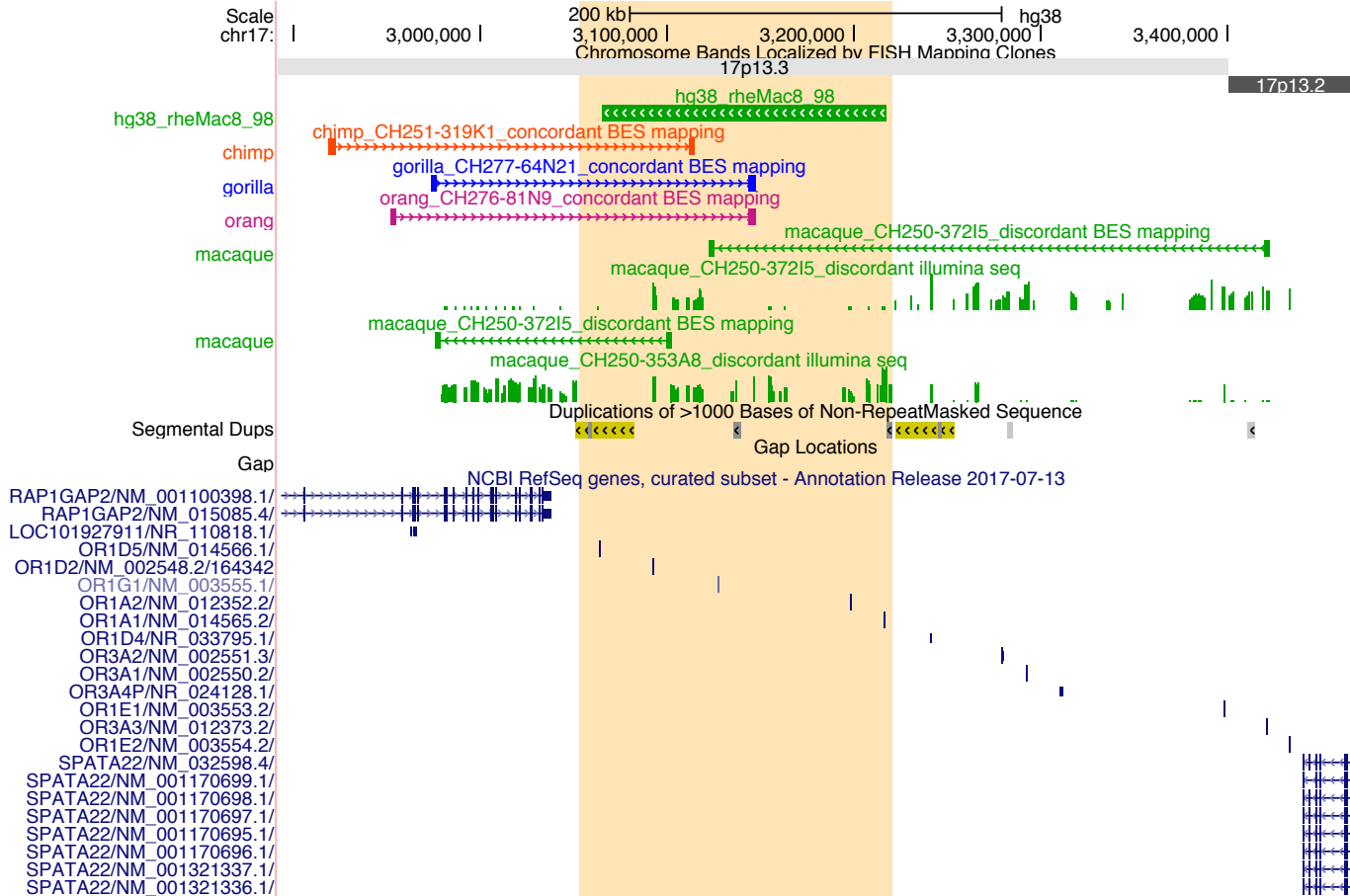


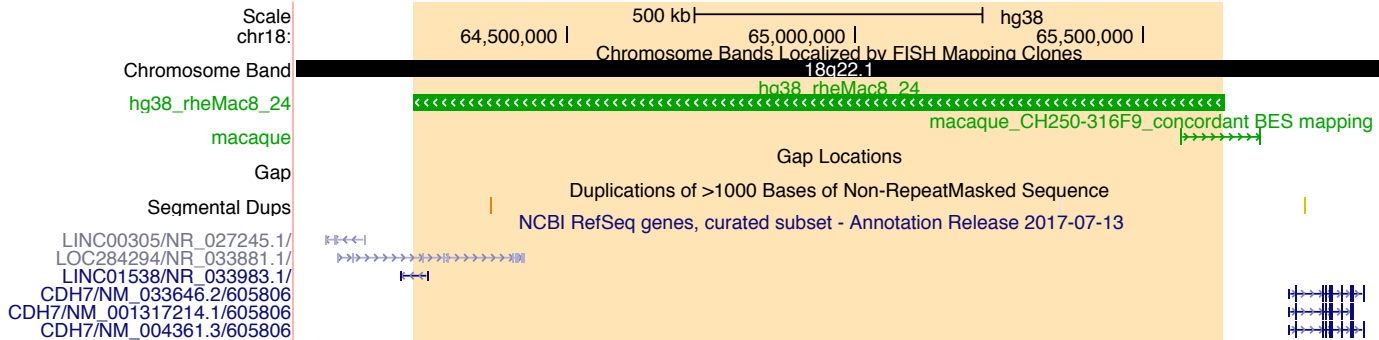


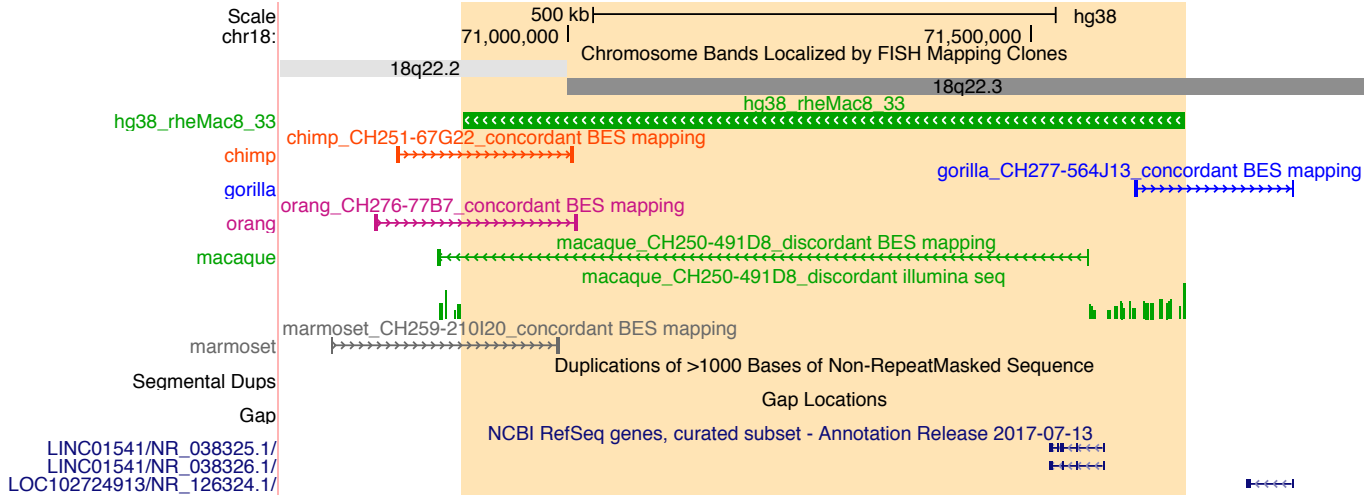


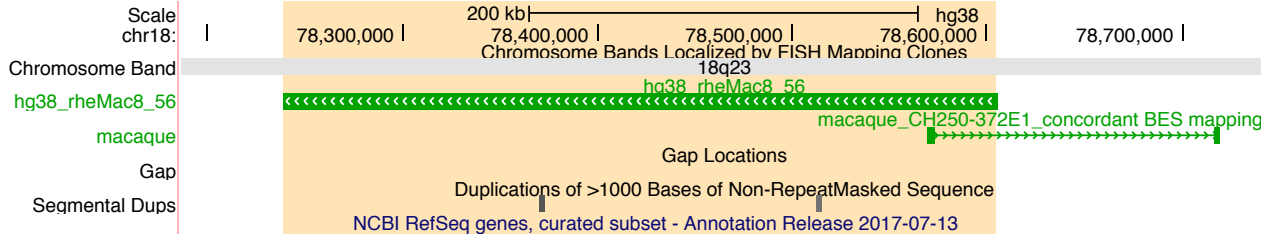












Scale chr18: | 68,200,000 | 200 kb | 68,300,000 | 68,400,000 | 68,500,000 | hg38 | 68,600,000 |

Chromosome Bands Localized by FISH Mapping Clones

18q22.1

hg38 rheMac8 69

hg38_rheMac8_69



chimp CH251-168D14 concordant BES mapping

chimp



chimp CH251-168D14 concordant illumina seq



gorilla

gorilla CH277-477O5 concordant BES mapping



gorilla CH277-477O5 concordant illumina seq



orang

orang CH276-35H16 concordant BES mapping



orang CH276-35H16 concordant illumina seq



macaque

macaque CH250-208C9 discordant BES mapping

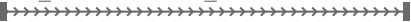


macaque CH250-208C9 discordant illumina seq



marmoset

marmoset CH259-409B17 concordant BES mapping



Gap Locations

Gap

Segmental Dups

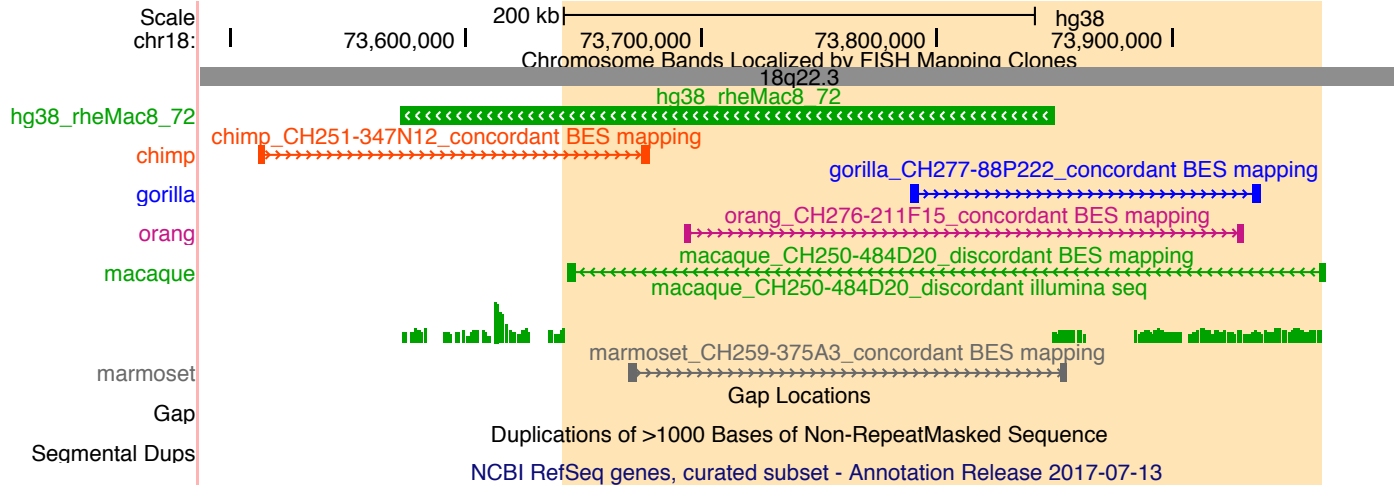
Duplications of >1000 Bases of Non-RepeatMasked Sequence



TMX3

NCBI RefSeq genes, curated subset - Annotation Release 2017-07-13





Scale chr18: | 5,650,000 | 5,700,000 | 5,750,000 | 5,800,000 | hg38 5,850,000 | 5,900,000 |

Chromosome Bands Localized by FISH Mapping Clones

18p11.31

hg38_rheMac8_103

hg38 rheMac8 103

chimp

chimp_CH251-526L2_concordant BES mapping
chimp_CH251-526L2_concordant illumina seq

gorilla

gorilla_CH277-383M2_concordant BES mapping
gorilla_CH277-383M2_concordant illumina seq

macaque

macaque_CH250-401F1_discordant BES mapping
macaque_CH250-401F1_discordant illumina seq

marmoset

marmoset_CH259-370O1_concordant BES mapping
marmoset_CH259-370O1_concordant illumina seq

Gap Locations

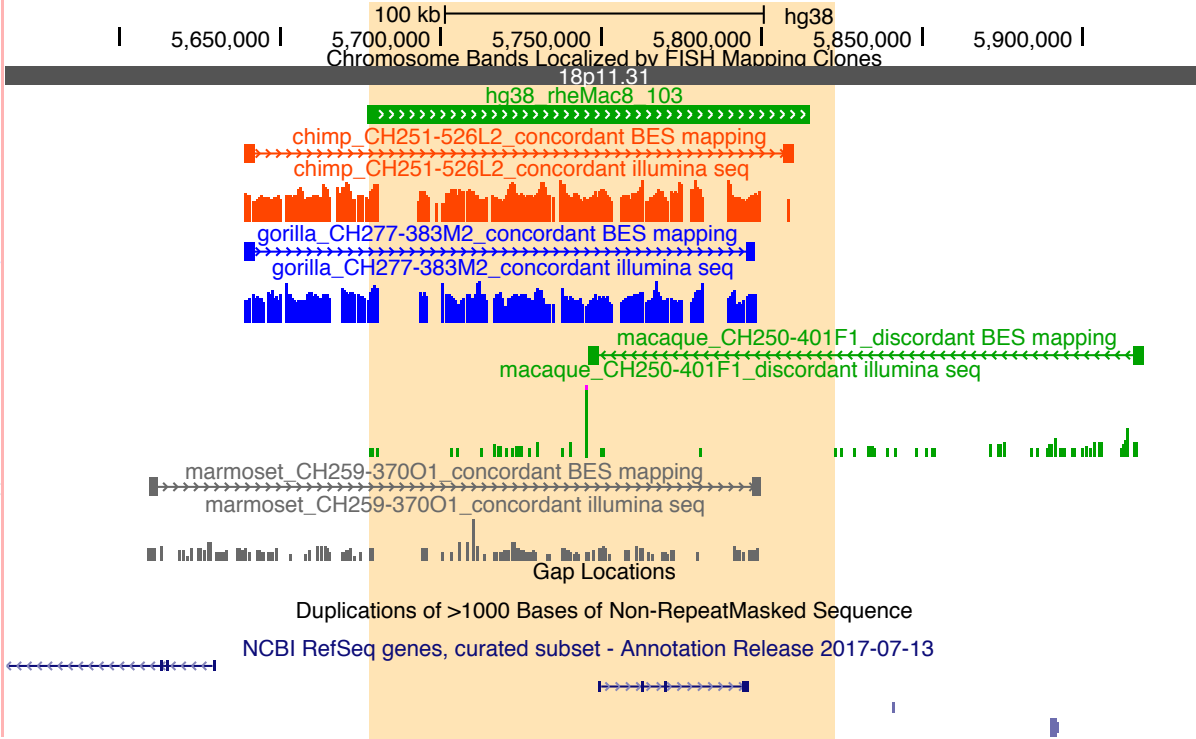
Gap

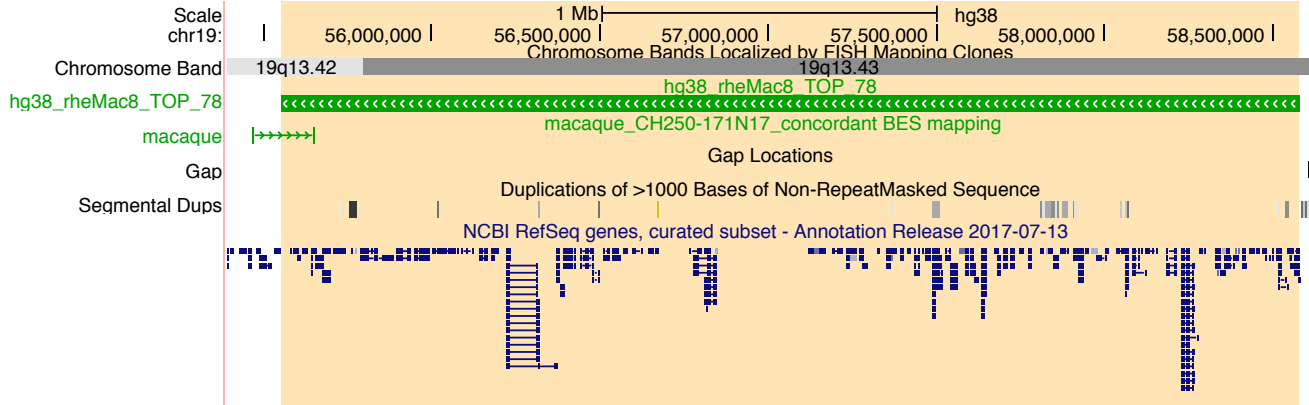
Duplications of >1000 Bases of Non-RepeatMasked Sequence

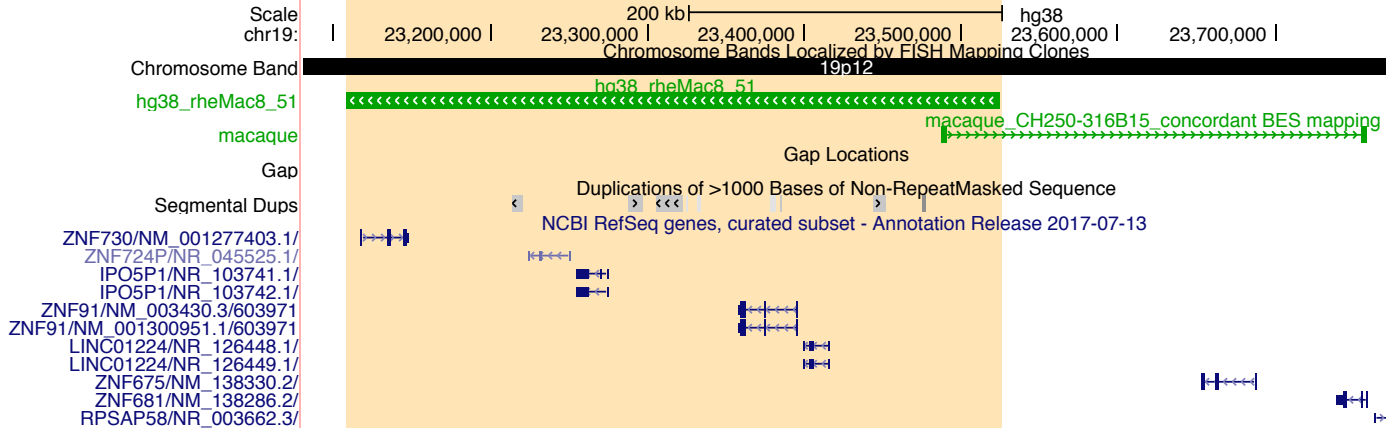
Segmental Dups

NCBI RefSeq genes, curated subset - Annotation Release 2017-07-13

EPB41L3
MIR3976HG
MIR3976
TMEM200C







Scale chr22: 32,100,000 | 100 kb | 32,150,000 | 32,200,000 | hg38 32,250,000 |

Chromosome Band 22q12.3

hg38_rheMac8_118

macaque macaque_CH250-530L10_concordant BES mapping

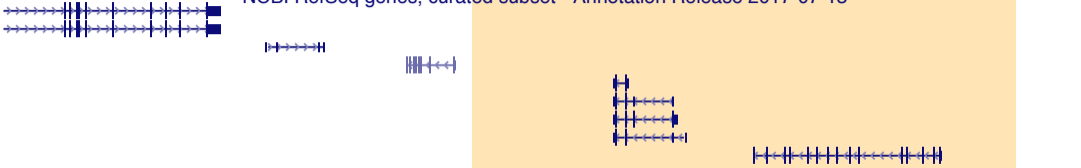
Gap Locations

Gap Segmental Dups

Duplications of >1000 Bases of Non-RepeatMasked Sequence

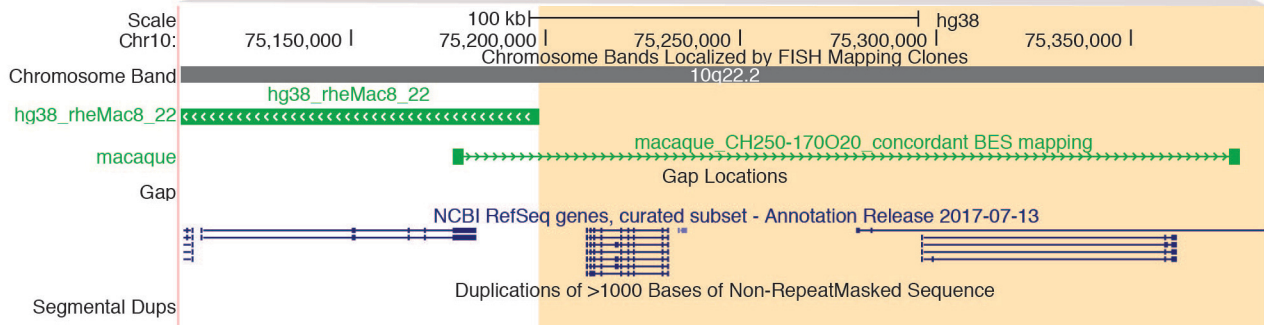
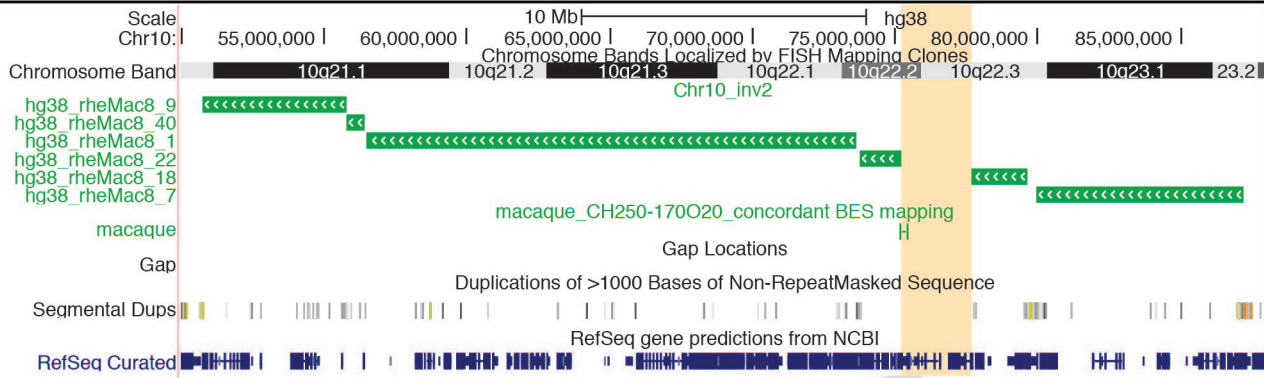
NCBI RefSeq genes, curated subset - Annotation Release 2017-07-13

SLC5A1/NM_000343.3/182380
SLC5A1/NM_001256314.1/182380
AP1B1P1/NR_040114.1/
C22orf42/NM_001010859.1/
RFPL2/NM_006605.3/605969
RFPL2/NM_001159546.1/605969
RFPL2/NM_001098527.2/605969
RFPL2/NM_001159545.1/605969
SLC5A4/NM_014227.2/



Supplemental Fig. S4. Experimental validation by BES pair mapping and Illumina sequencing of BAC clones.

UCSC Genome Browser views of all the inversion calls tested by Illumina sequencing and/or BES pair mapping. All BAC clones spanning the breakpoints of the predicted regions are shown as colored segments. Illumina sequenced discordant clones mapping at the breakpoints of the inversions appear as discontinuous. BES pair mapping of discordant clones supporting the inversion have end pairs that map abnormally far apart and have ends that are incorrectly oriented when mapped to the human reference genome sequence. Segmental duplications, sequence gaps and RefSeq curated genes are also shown for each event.



Supplemental Fig. S5. Example of a misassembled region in the macaque genome.

The cytogenetic inversion on human Chromosome 10 is 36.5 Mb in size and was detected as six separate smaller inversion calls. Nevertheless, the calls hg38_rheMac8_22 and hg38_rheMac8_18 are interrupted by an interval of 2.5 Mb of sequence in direct orientation. We investigated the orientation of this region by BES pair mapping. Clone CH250-170O20, spanning the proximal breakpoint of this region, demonstrates that it is in the same orientation of its flanking sequences and therefore represents an assembly error.