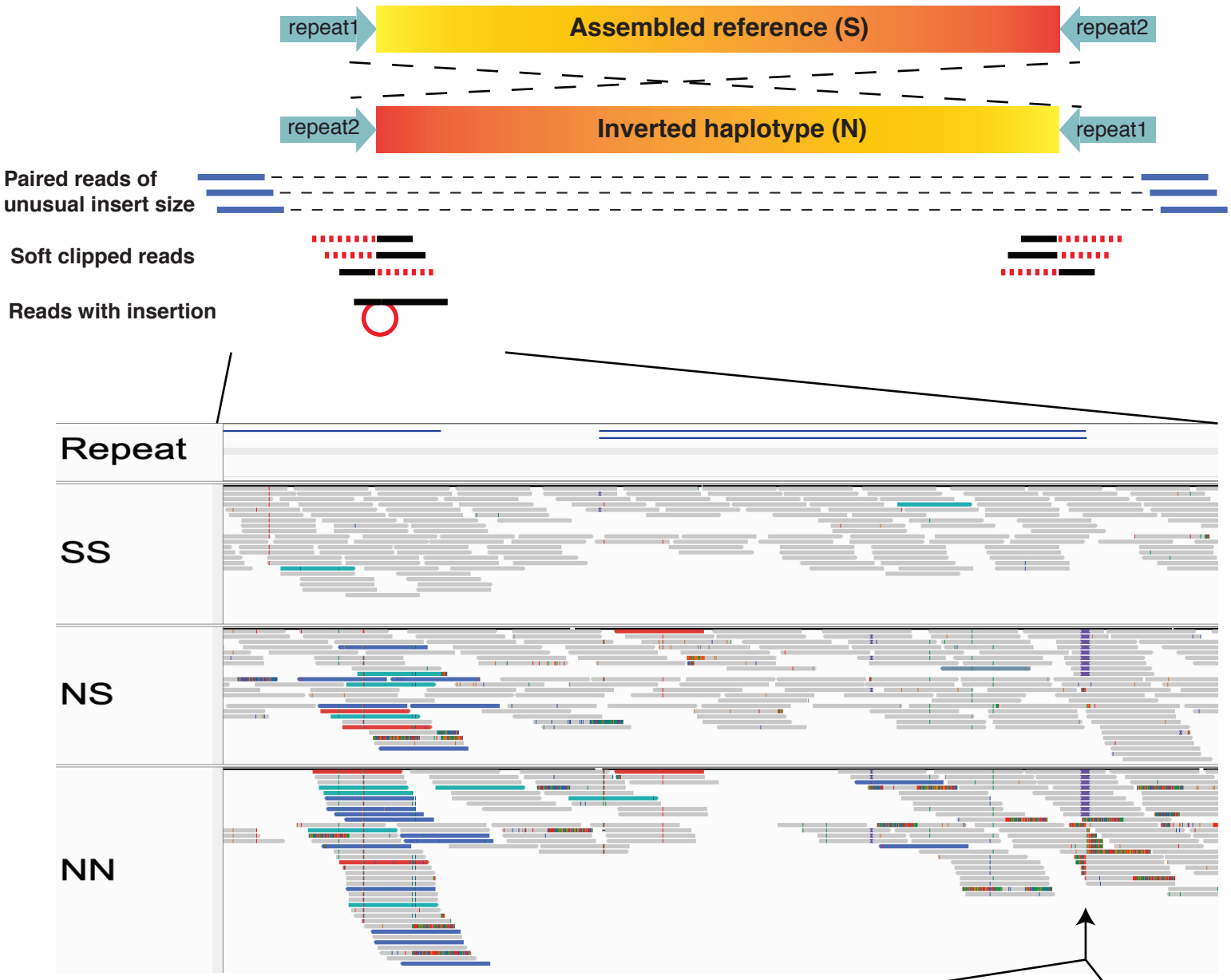
A)


repeat1
Assembled reference (S)
repeat2
repeat2
Inverted haplotype (N)
repeat1
Paired reads of unusual insert size
Soft clipped reads
Reads with insertion
Repeat
SS
NS
NN

B)

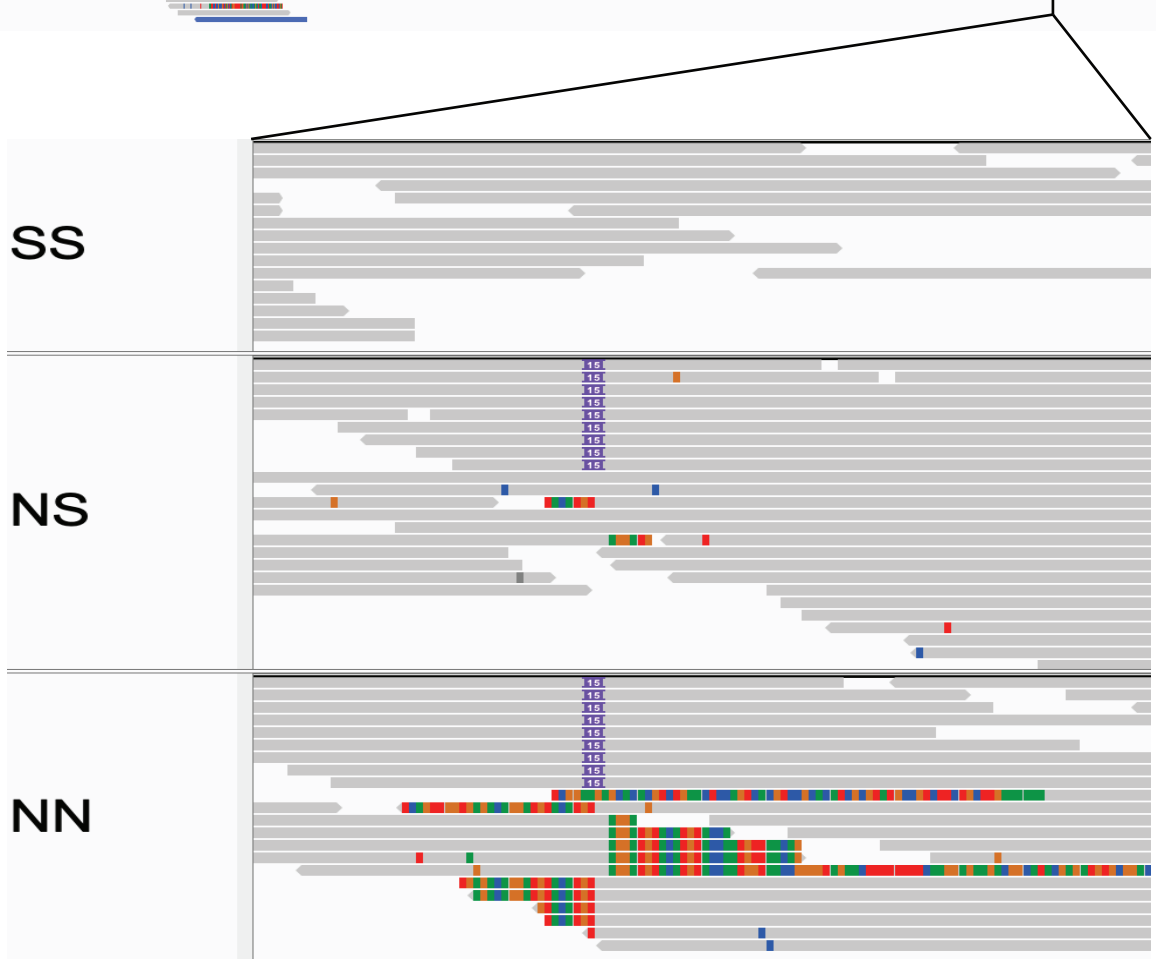

SS
NS
NN

Figure S2: Schematics of sequence structure and chimeric read alignment at the Chr 12 inversion. (A*)* Schematics of sequence structure at the Chr 12 inversion. An 815 bp inverted repeat is located upstream of the 7.8 Mb inversion, followed by its reverse complement downstream. Short-read alignments from individuals homozygous (NN) for the Northern haplotype show three patterns around the breakpoints: insert size of paired reads of about 8 Mb consistent with the presence of a 7.8 Mb inversion; Reads are soft-clipped (red dashed line) at the same position; Unclipped reads contain a small insertion (red loop) which has an identical sequence as the soft clipped reads (red dashed line). (*B)* A zoomed-in view of read alignment at the putative breakpoint in IGV. Reads from SS individuals perfectly align to the breakpoint region as the reference assembly is based on the S haplotype. The reads from NN individuals are either soft clipped at the same base or bear a 15 bp insertion at the clipped site and it has the same sequence as the clipped bases. About half of the reads from heterozygous individuals (NS) are properly aligned like reads from SS individuals while the other half behave as those from NN individuals.