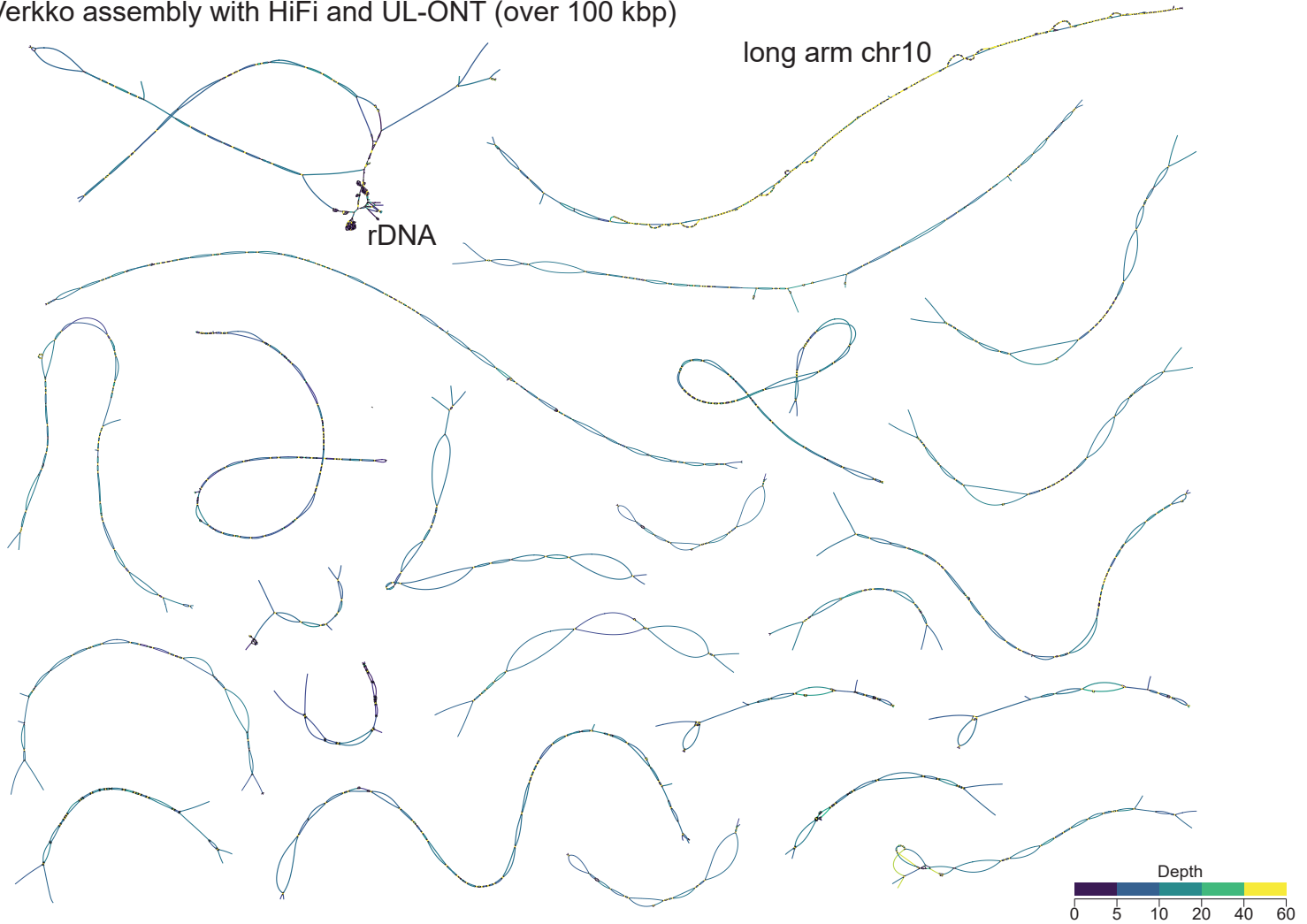
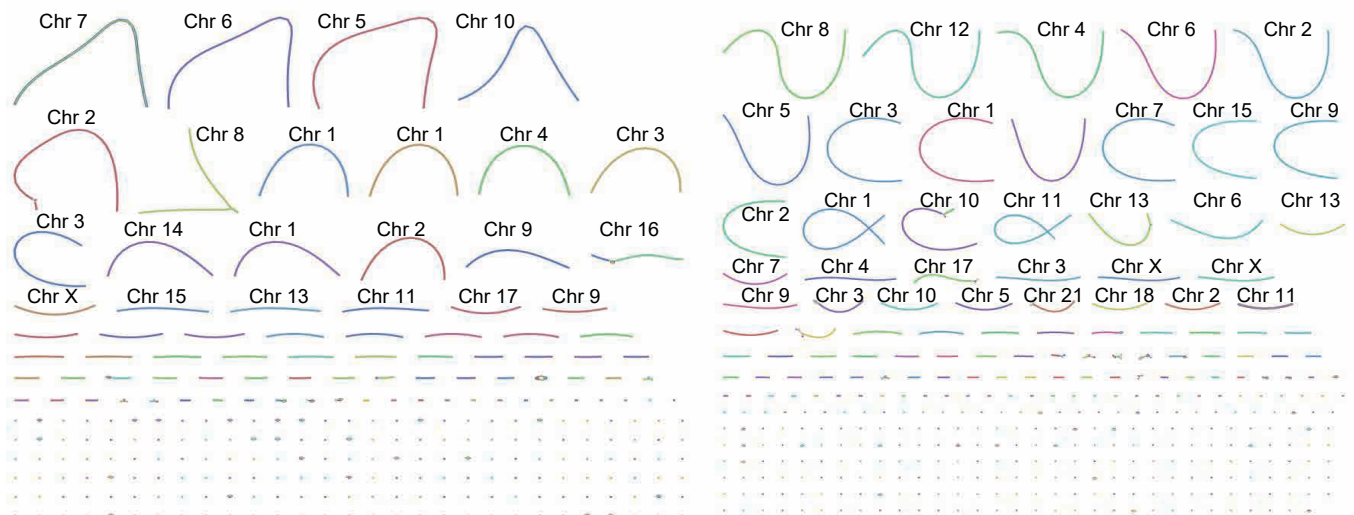
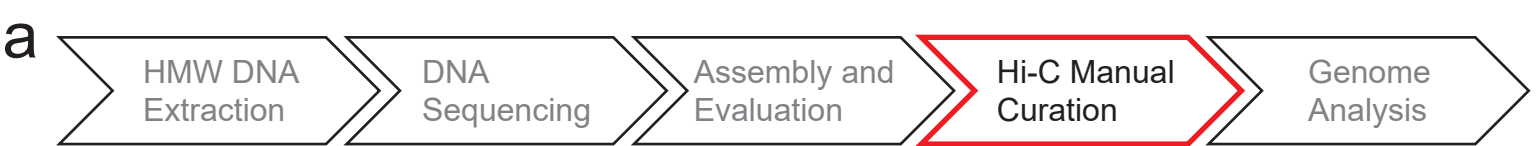


b Verkko assembly with HiFi and UL-ONT (over 100 kbp)

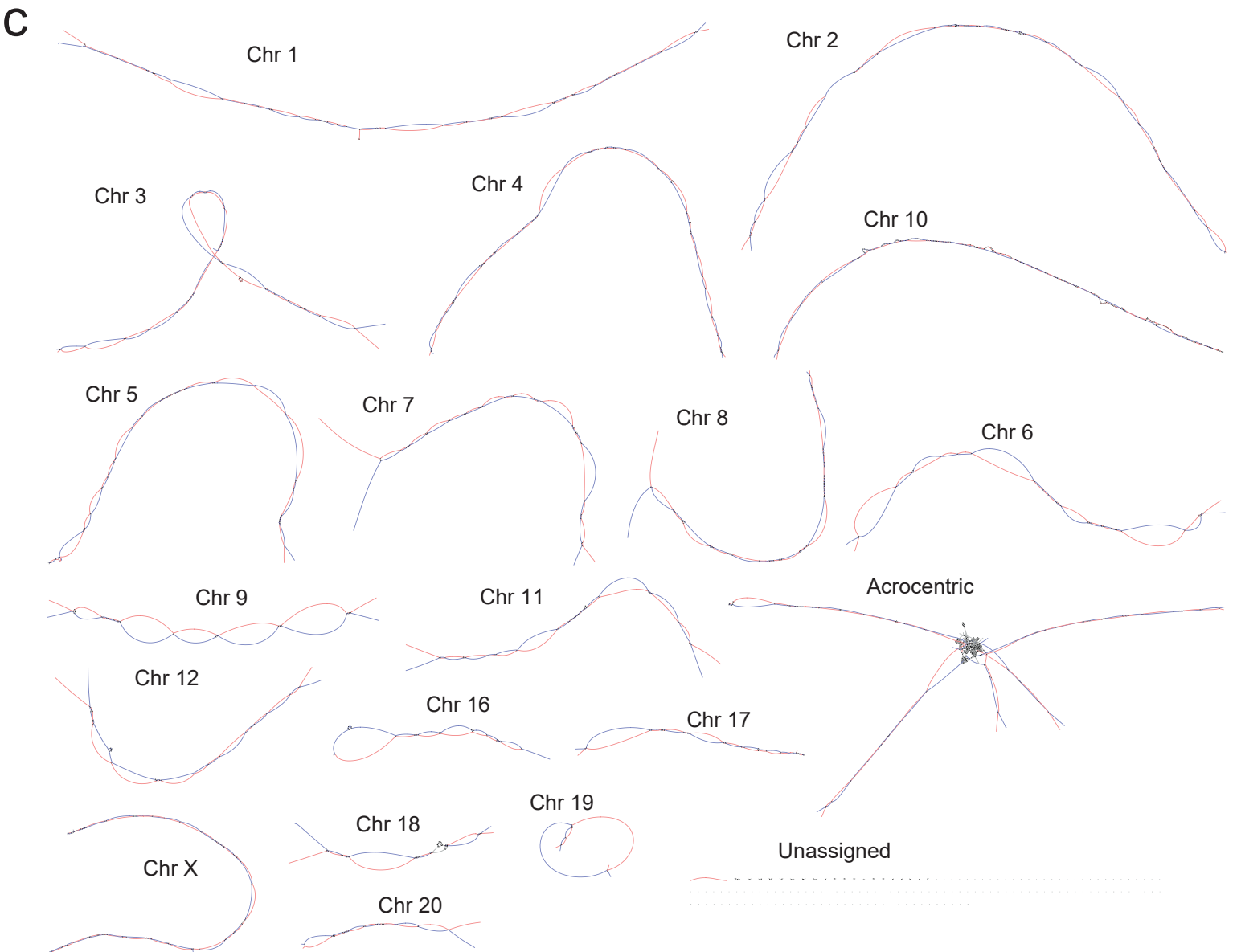
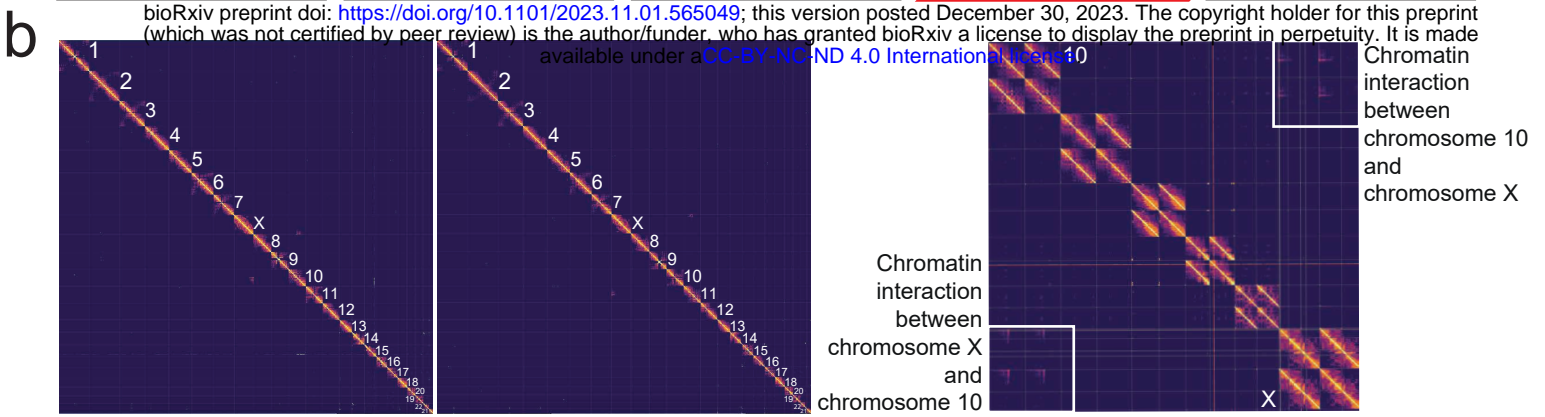


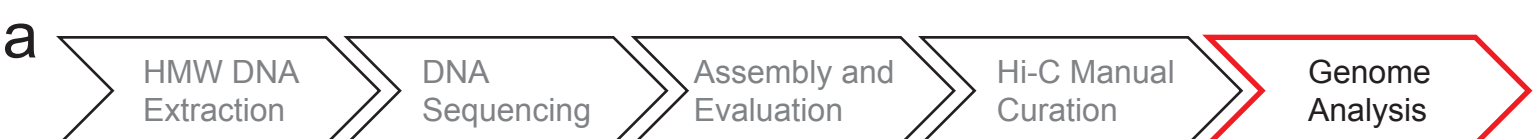
c Hifiasm partially-phased assembly



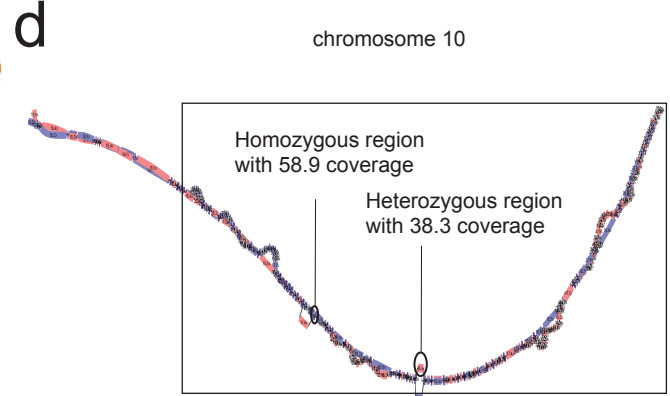
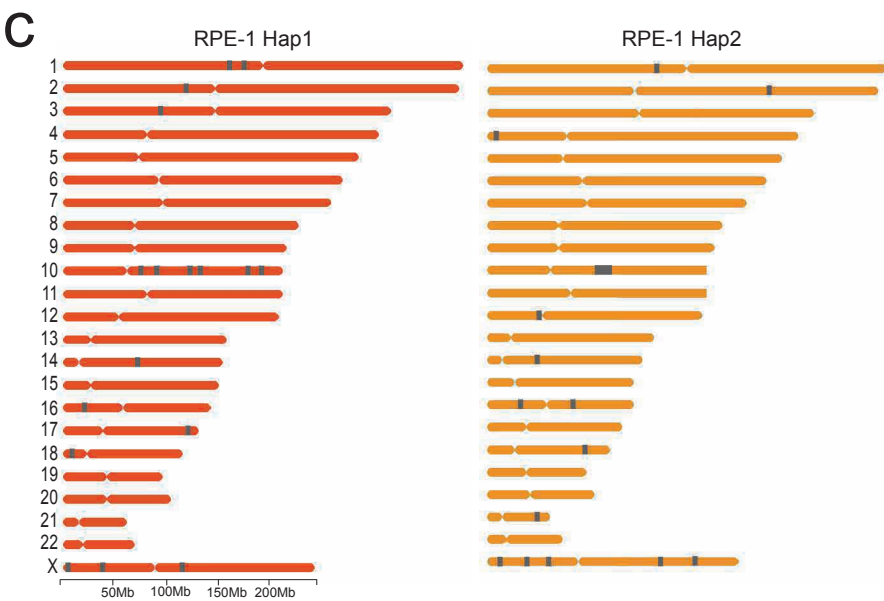
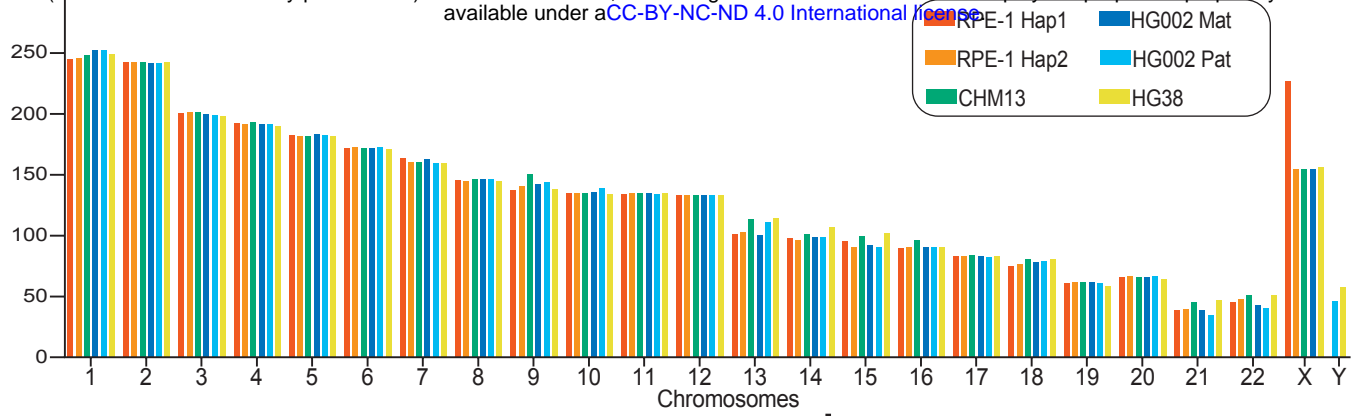


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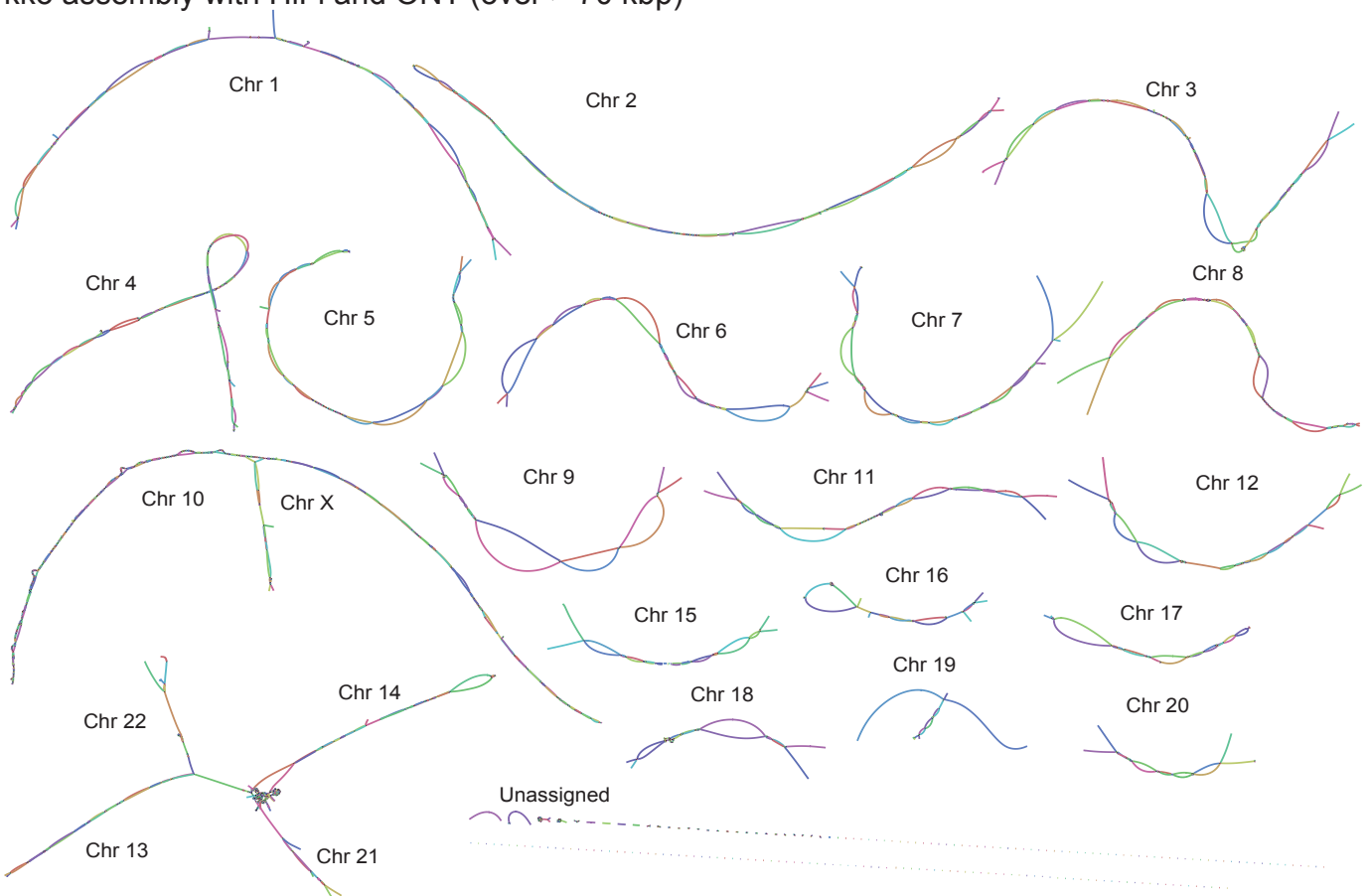


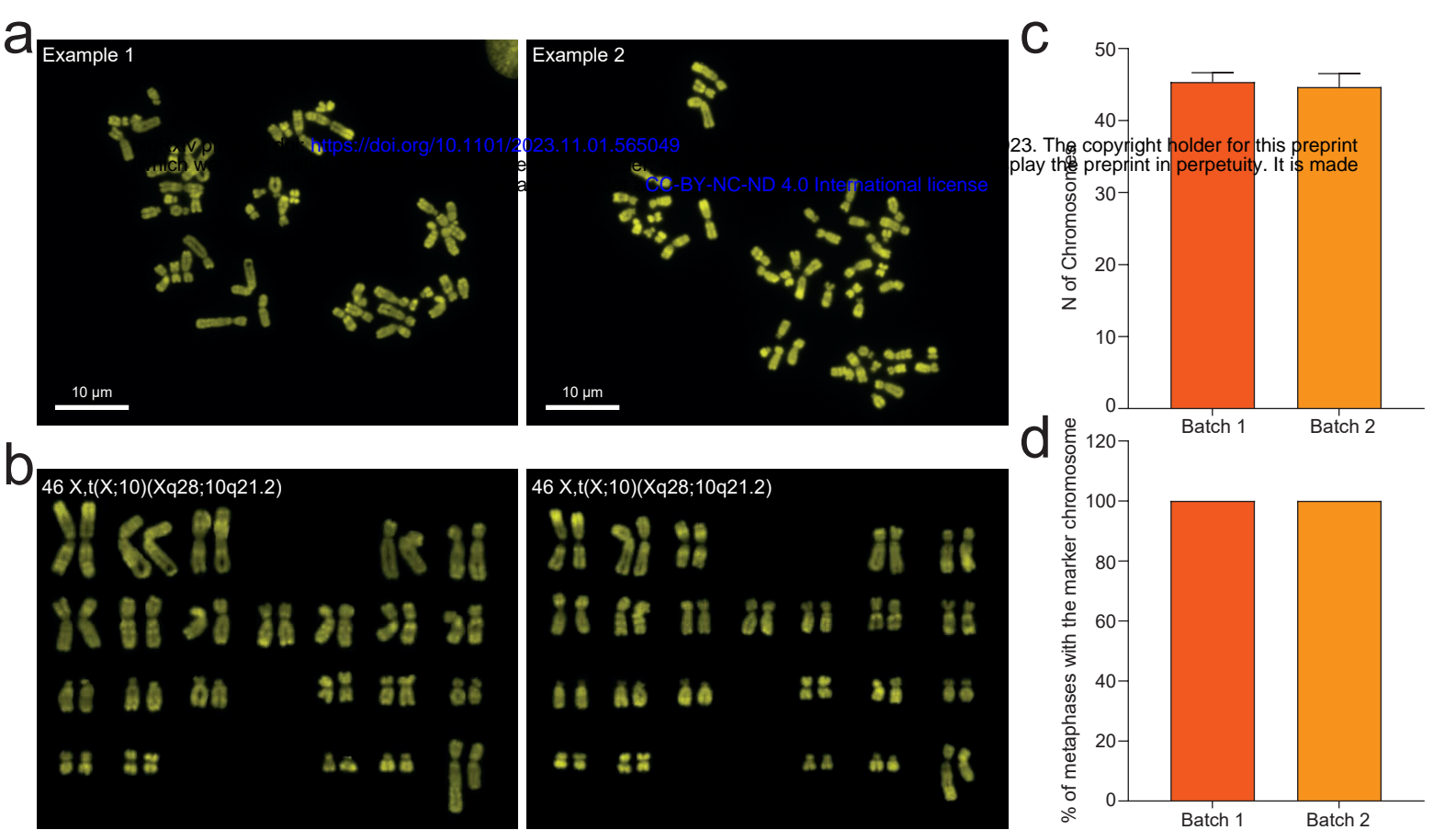


b bioRxiv preprint doi: <https://doi.org/10.1101/2023.11.01.565049>; this version posted December 30, 2023. The copyright holder for this preprint (which was not certified by peer review) is the author/funder, who has granted bioRxiv a license to display the preprint in perpetuity. It is made available under a [CC-BY-NC-ND 4.0 International license](https://creativecommons.org/licenses/by-nc-nd/4.0/).







e Verkko assembly with HiFi and ONT (over > 70 kbp)

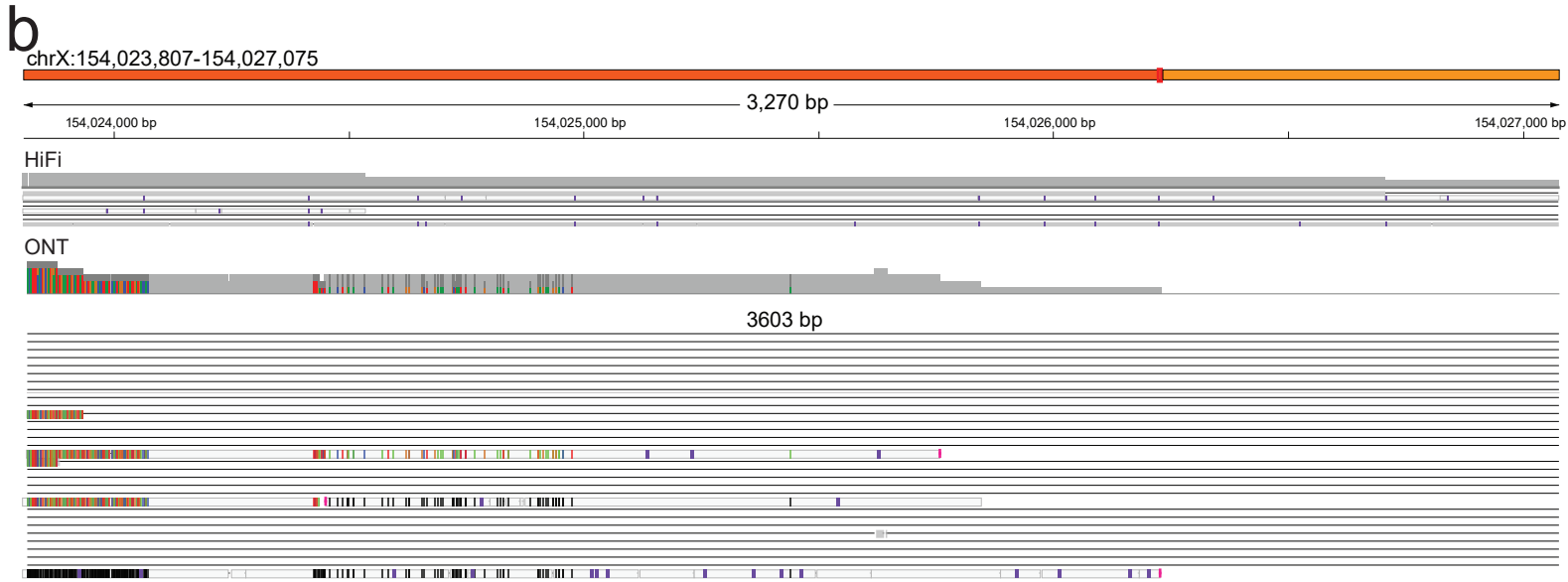
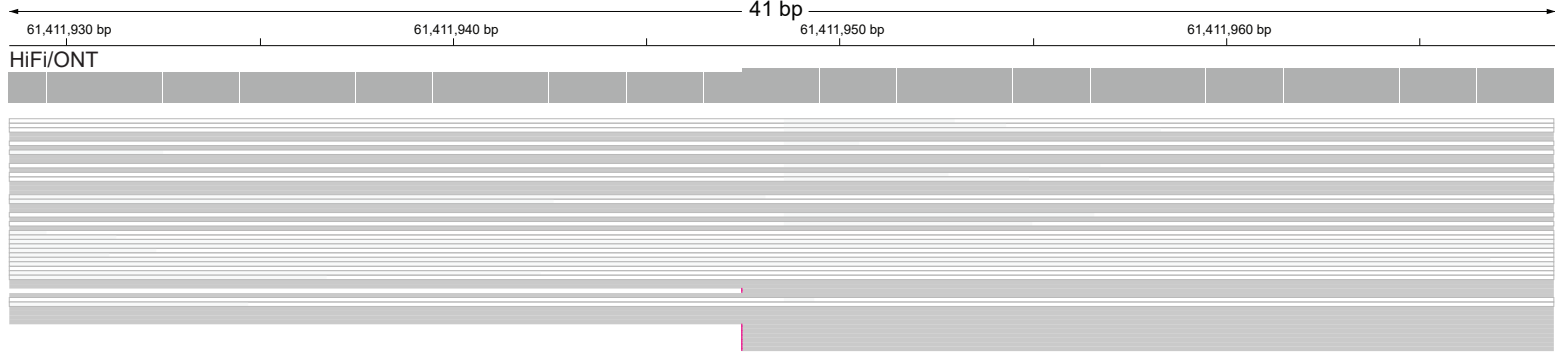




Translocation localization

 = Mapq 10-60, supplementary alignments chrX hap1
 = 3603 bp deletion
 chr10 Hap2
 chrX Hap1

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chr 1

chr 3

chr 7

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Hap1

1

2

Hap2

chr 8

chr9

chr 10

chr 12

chr13

chr 15

chr 17

chr 18

chr 19

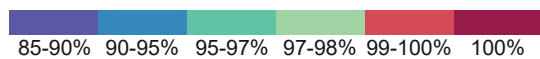
chr20

chr 21

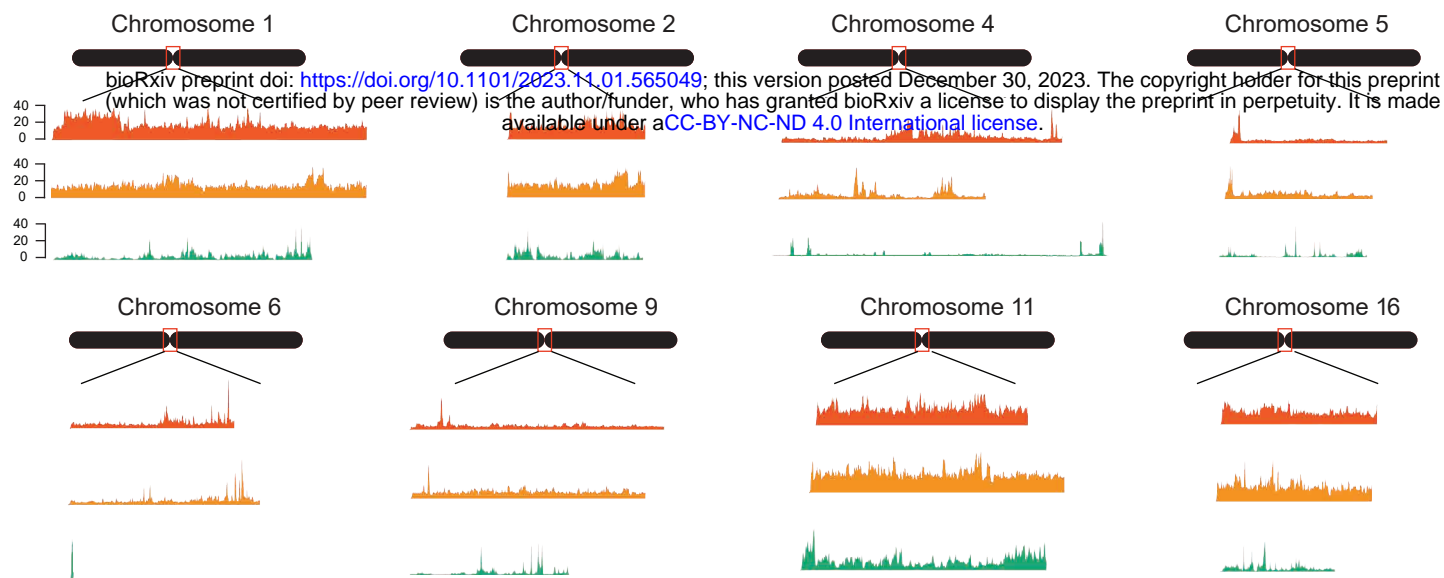
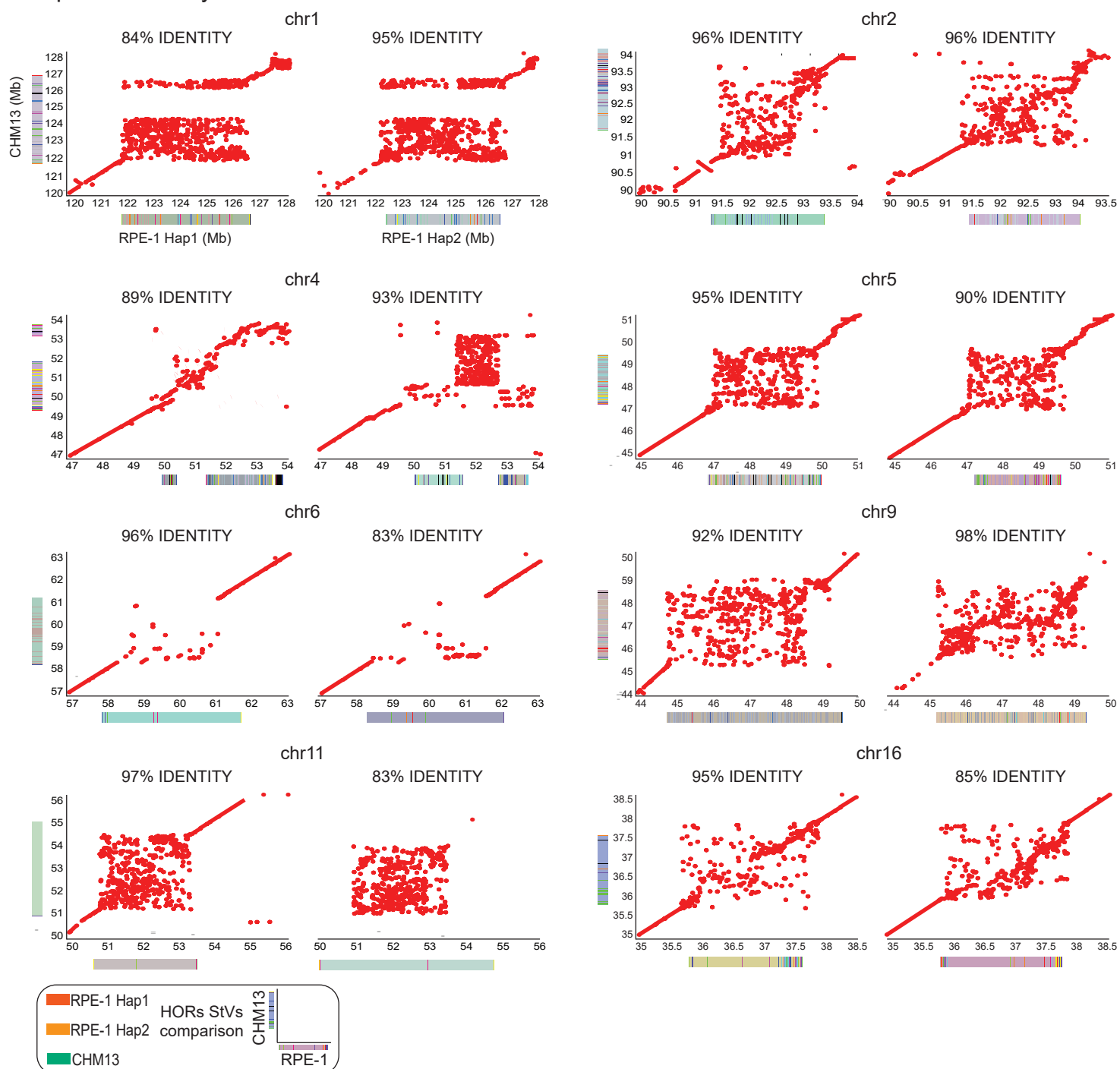
chr 22

1=LHOR organization in Super Families (SF0-3)

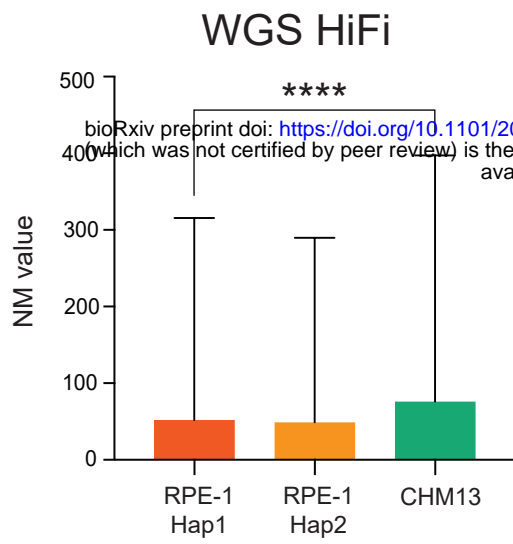
2=Specific variants for each haplotype



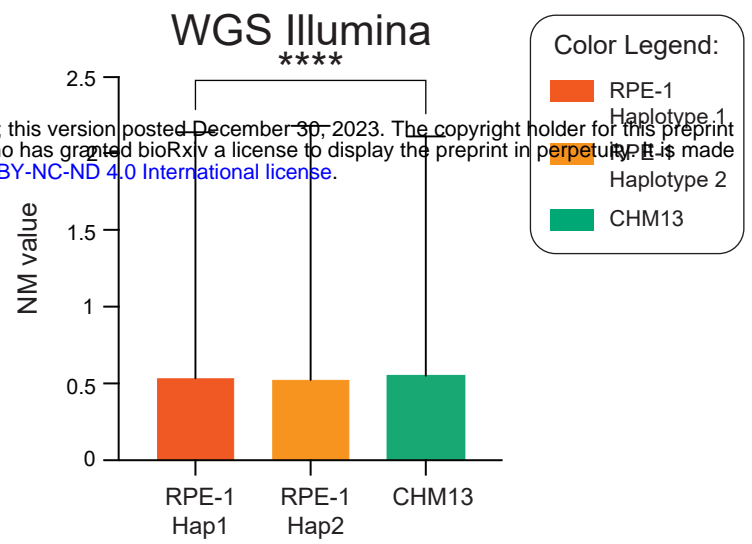
85-90% 90-95% 95-97% 97-98% 99-100% 100%

a**IHORs Coverage****b****Sequence Identity**

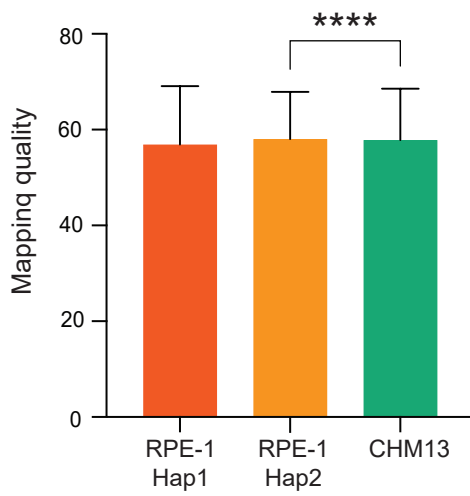
a



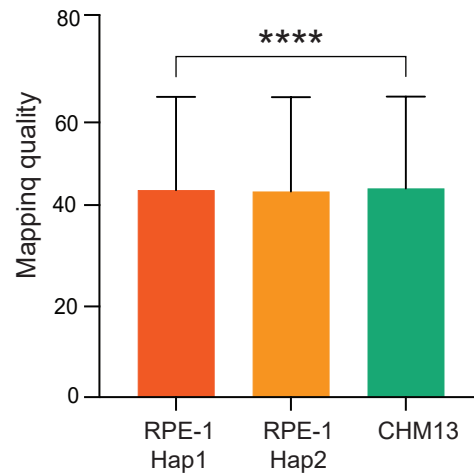
b



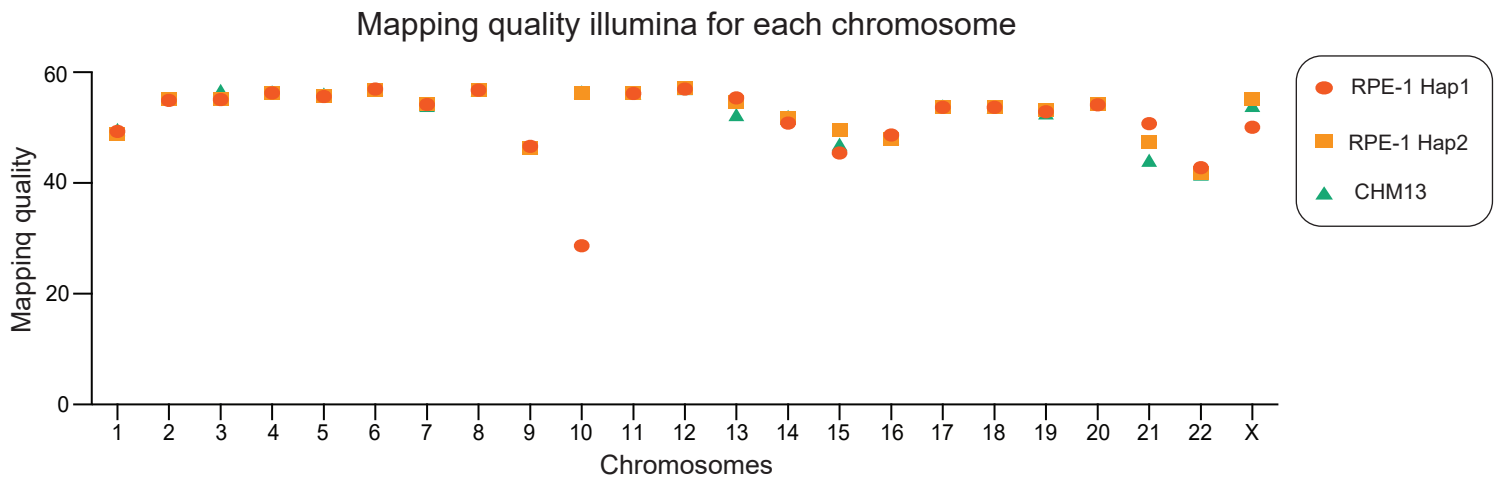
c



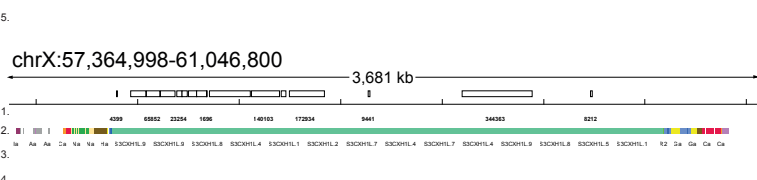
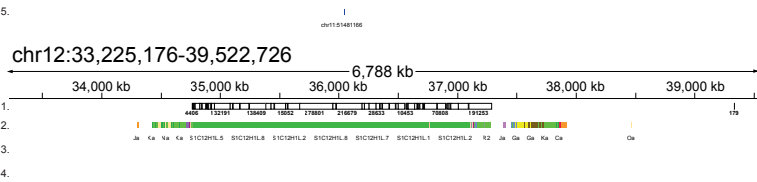
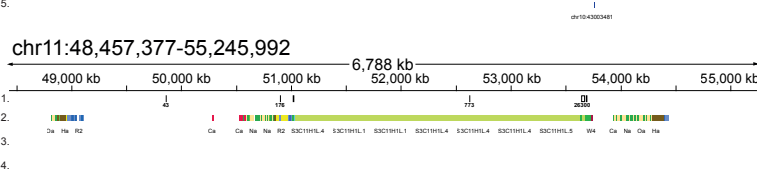
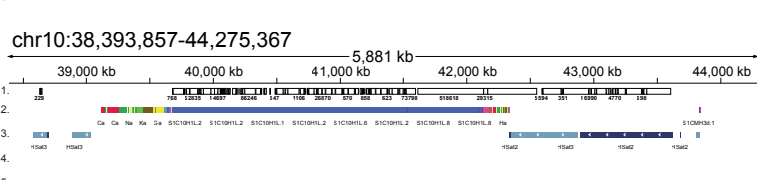
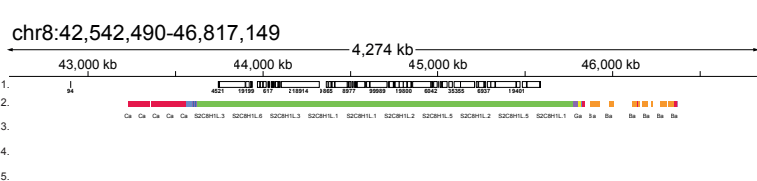
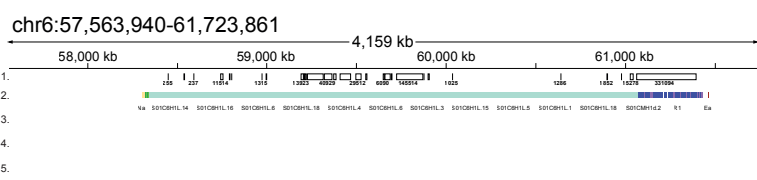
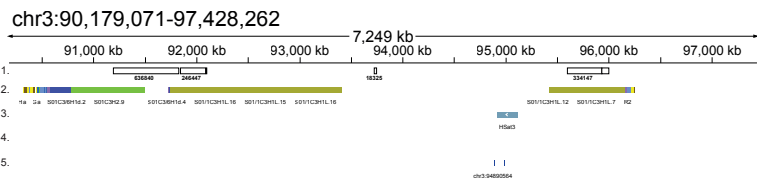
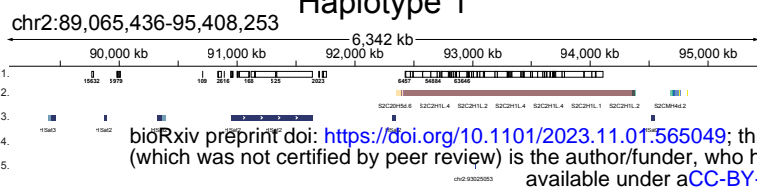
d



e



Haplotype 1



1. Highly Diverged Region annotation
2. Live High Order Repeat annotation
3. Human satellite annotation
4. Assembly regional errors
5. Assembly regional heterozygous loci

Haplotype 2

