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Equine recombination map updated to EquCab3.0

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We recently investigated fine-scale recombination rate variability in the domestic horse¹ using high-density genotyping data from the MNEc2M SNP array² and the EquCab2 reference genome assembly³. Recent completion of EquCab3⁴ has resulted in an overall higher-quality assembly which includes inversions and translocations relative to EquCab2. Further, thousands of SNPs on the MNEc2M array, previously mapped to unplaced contigs in EquCab2, now map to equine chromosomes (ECA) 1-31 and ECAX in EquCab3⁵. The purpose of this report is to update the domestic horse recombination map to EquCab3 coordinates to ensure its continued usefulness to the equine genome research community.

Our dataset consists of previously generated SNP genotypes of 485 horses from 32 breeds². Genomic coordinates for these data were originally based on the EquCab2 reference build³; therefore, we performed probe-based remapping to EquCab3⁵, producing a final set of 1,820,349 biallelic SNP genotypes, which we phased using BEAGLE⁶. We generated recombination maps as described previously¹. Briefly, we sampled 40 haplotypes 100 times per chromosome from the dataset described above and estimated population recombination rates ($4N_e r$) using *LDhat*⁷, taking the average across random samples. For 12 individual breeds (Arabian, Belgian, Franches-Montagnes, French Trotter, Icelandic, Lusitano, Maremmano, Morgan, Quarter Horse, Standardbred, Thoroughbred, and Welsh Pony), we created breed-specific maps following the same procedure. Using these recombination rate estimates, we predicted recombination hotspots with *LDhot*⁸. Finally, we converted recombination rates from $4N_e r$ to centimorgans (cM) with effective population size estimates generated previously¹. More details are provided in the Supplemental Methods.

The equine genome recombination map updated to EquCab3 spans 2,794.81 centimorgans (cM) over 2.41 Gb. As a result, the average genome-wide recombination rate is 1.16 cM/Mb. This is lower than our previous estimate of 1.24 cM/Mb¹. Table 1 and Supplemental Figure S1 contain a summary of chromosomal recombination rates and fine-scale rates by chromosome, respectively. In all cases, the estimated rate across individual chromosomes is lower in EquCab3 than in EquCab2¹ (Supplemental Table S1). The median width of predicted recombination hotspot regions is 3.1 kb, with an overall range of 1-10 kb. Hotspots

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Data Availability

Recombination maps and hotspots are available at <https://osf.io/e72ah>.

now account for approximately 155.3 Mb, or 6% of the equine genome, and 1,416.9 cM, or 50.6% of the genetic map length, both of which are reductions from previous estimates. In predicted hotspot regions, the mean recombination rate is 9.1 cM/Mb, roughly 8 times the genome-wide average. Breed-specific maps are summarized in Supplemental Table S2. Overall, differences between the updated recombination map and our previous map appear to reflect improvements in the equine genome assembly and provide an improved means for investigating the basis of equine genome structure and function.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

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Table 1.

Physical distance, genetic distance, average recombination rate, and hotspot count by chromosome in the updated recombination map.

| | Genetic length (cM) | Physical length (Mb) | Mean rate (cM/Mb) | Hotspots |
|---------------|---------------------|----------------------|-------------------|----------|
| Genome | 2,794.81 | 2,408.75 | 1.16 | 42,243 |
| ECA1 | 190.98 | 188.25 | 1.01 | 2,962 |
| ECA2 | 134.22 | 121.34 | 1.11 | 1,969 |
| ECA3 | 129.48 | 121.35 | 1.07 | 1,987 |
| ECA4 | 112.60 | 109.46 | 1.03 | 1,843 |
| ECA5 | 102.97 | 96.75 | 1.06 | 1,737 |
| ECA6 | 94.92 | 87.22 | 1.09 | 1,594 |
| ECA7 | 113.78 | 100.78 | 1.13 | 1,764 |
| ECA8 | 101.07 | 97.55 | 1.04 | 1,544 |
| ECA9 | 91.71 | 85.75 | 1.07 | 1,435 |
| ECA10 | 105.89 | 85.15 | 1.24 | 1,505 |
| ECA11 | 91.71 | 61.66 | 1.49 | 1,088 |
| ECA12 | 71.06 | 36.98 | 1.92 | 858 |
| ECA13 | 57.88 | 43.78 | 1.32 | 902 |
| ECA14 | 99.99 | 94.59 | 1.06 | 1,547 |
| ECA15 | 95.74 | 92.85 | 1.03 | 1,601 |
| ECA16 | 94.46 | 88.94 | 1.06 | 1,497 |
| ECA17 | 88.09 | 80.71 | 1.09 | 1,463 |
| ECA18 | 84.47 | 82.62 | 1.02 | 1,414 |
| ECA19 | 75.39 | 62.67 | 1.20 | 1,106 |
| ECA20 | 92.01 | 65.22 | 1.41 | 1,281 |
| ECA21 | 68.05 | 58.94 | 1.15 | 1,051 |
| ECA22 | 53.25 | 50.92 | 1.05 | 954 |
| ECA23 | 57.68 | 55.56 | 1.04 | 917 |
| ECA24 | 57.52 | 48.27 | 1.19 | 842 |
| ECA25 | 46.27 | 40.27 | 1.15 | 695 |
| ECA26 | 62.31 | 43.12 | 1.45 | 741 |
| ECA27 | 46.72 | 40.25 | 1.16 | 755 |
| ECA28 | 54.21 | 47.34 | 1.15 | 828 |
| ECA29 | 51.13 | 34.77 | 1.47 | 705 |
| ECA30 | 42.94 | 31.39 | 1.37 | 597 |
| ECA31 | 34.27 | 26.00 | 1.32 | 1,079 |
| ECAX | 192.04 | 128.20 | 1.50 | 1,982 |