The influence of demography and recombination on the homozygosity landscape in the poultry genome

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1. Introduction

Inbreeding has been recognised as one of the main driving forces of extinction in small populations, along with demographic stochasticity and reduced genetic diversity. The increased homozygosity observed in inbred populations may result in redistributed genetic variation within and between populations, lowered population mean of fitness-related traits, and higher incidence of homozygous recessive defects. Inbred populations suffer from the effects of parental relatedness because matings between consanguineous cause the inheritance of haplotypes that, if traced back in time, belong to the same common ancestor (IBD). Although the inheritance of IBD segments results in homozygous stretches along the genome, these regions of homozygosity (ROHs) are expected to not be uniformly distributed across the genome, but to be more prevalent in certain regions, ROH islands or hotspots, than others, ROH deserts or coldspots. The existence of these ROH hotspots and coldspots can partly be explained by stochasticity in recombination events across the genome or variation in the effects of demographic processes that influence genetic diversity. Moreover, the size and position in the genome of ROHs are expected to correlate with specific genomic features, including the recombination rate. Here we used next-generation sequencing data for an in-depth analysis of the genomic ROH patterns of 37 traditional Dutch chicken breeds, comprising large fowls, true bantams, and bantamised forms, and four living wild species of *Gallus*. The distinct demographic and management history of Dutch chicken breed make them a good model for investigating the effects of demography, and thus inbreeding, in marginalised populations.

2. Approach

ROHs were inferred in each individual using the sliding window approach implemented by Bosse et al. (2012). Although inferring ROHs from sequence data is particluarly challenging, the approach here implemented wants to overcome the challenges and pitfalls of current tools developed for SNP arrays, which are not suitable for detecting ROHs from sequence data. Briefly, we defined an ROH as a genomic region of at least 10 Kbps where the number of SNPs per bin (or SNP count) was less than 0.25 the whole-genome nucleotide diversity, and if at least 10 consecutive bins showed a total SNP average lower that the total genomic average. Local assembly or alignment errors were avoided by relaxing the threshold for individual bins within a candidate homozygous stretch. Recombination rate was calculated from the chicken linkage map and averaged over all markers in each ROH.

3. Results

We found an average of 280.8 ($\pm$ 87.78) ROHs/genome, with an average size of 1359.80 ($\pm$ 584.3) Kbps and an average total size of 372.73 Mbps ($\pm$ 139.27). Average ROH size and average number of ROHs in the genome did not significantly differ in the 4 groups comprising the traditional breeds, compared to the wild types, which showed a more heterogenenous ROH profile, along with higher average ROH size (2360 Kbps) and lower ROHs number (63.75). At population level, we observed remarkable differences between breeds and between *in-situ* and gene bank collections. Samples from the gene bank showed an overall lower number of ROHs compared to the relative *in-situ* populations, though the total length and average ROH size differed with the breed. The Eikenburger bantam was the most inbred breed in our dataset. In fact, although the total number of ROHs in the genome was relatively low, homozygous stretches were predominately medium-long, leading to roughly 600 Mbps of genome covered by ROHs. Recombination rate averaged per chromsome across all populations strongly correlated with the ROH size (-0.84, p-value 1.697-e08).

4. Discussion

We studied the influence of demography and recombination on the genomic homozygosity in order to better understand the effects of inbreeding. Size and number of ROHs were in agreement with the population demographic history reported in our previous study, confirming the impact of bottlenecks, genetic drift, and consanguineous matings on the genomic ROH profile. Moreover, we showed that both demography and recombination play a major role in determing the ROH landscape. Our findings show that the approach here used to detect ROHs from whole-genome sequence data can easly be applied to other livestock species, thus exceeding species boundaries. Moreover, compared to current SNP array tools, ROHs of different lengths were equally detected, thus providing a better and complete picture of the past and current demographic history of populations.

References

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